GENETIC AND EPIGENETIC PROFILES OF ELDERLY AML

ACUTE MYELOID LEUKEMIA IN THE ELDERLY IS CHARACTERIZED BY A DISTINCT GENETIC AND EPIGENETIC LANDSCAPE

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HUMBOLDT-UNIVERSITÄT ZU BERLIN

We stumble on a mystery and embrace it.
We investigate and we doubt, but it would be
ignorance to believe the inexplicable is impossible.
In the end, our reward is a story worth telling.
One that not only contributes to knowledge but also transforms us.

Patricia Silva

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DEDICATION

I am dedicating this thesis firstly to my beloved husband, António Nascimento, who always gave me his unwavering support and positivism.

Then to my parents (Luis and Adelina Silva) and my brother (André Silva) who were essential through the years to get here. My parents especially, for they are my heroes and that will always mean everything to me. They gave me more than you can ask from any parent.

Also to my supervisor, Claudia Baldus, for I was very fortunate to have found her. I am grateful to her for providing the right balance of leadership and freedom, paramount to the realization of this work.

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I personally have to thank all the people that were direct and indirectly responsible for this achievement.

To all members of the lab, a big thank you for your everyday support and helpful suggestions. I will be forever thankful to all of you for providing the best environment to work in. I want to thank Liliana Mochmann for providing more than scientific discussions, for the friendship that made my life happier.

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AUTHORSHIP DECLARATION

The findings I present in this publication were obtained and written by me and were previously published in the *Leukemia* journal (NPG®) where I am the first author[1], namely: Silva, P. *et al.* Acute myeloid leukemia in the elderly is characterized by a distinct genetic and epigenetic landscape. *Leukemia* **31,** 1640–1644 (2017). Therefore most chapters and pictures were adapted from my previous manuscript, some were extended and some were added new. Statements from my previous abstract in *Blood* journal and respective oral presentation in the American Society of Hematology meeting 2015[2] might also be present, namely from: Silva, P. *et al.* Acute Myeloid Leukemia in the Elderly Is Characterized By a Distinct Genetic Landscape. *Blood* **126,** (2015).

A research like ours surely had to be a joint effort. We all know collaborations between researchers and fellow scientists are essential to achieve important knowledge in such an interdisciplinary area as translational medicine. Therefore, although I was involved in all the parts of the project from design to experiments (where I prepared libraries, implemented strategies/workflows, performed computational and statistical analysis and confirmed mutations) I cannot forget others. The physicians that treated patients, collected samples and information and the technicians that treated the samples and isolated gDNA. So I would like to leave a more detailed account of contributions from the several collaborators to this work (in the next chapter).

I hereby declare that I completed the doctoral thesis independently based on the stated resources and aids. I have not applied for a doctoral degree elsewhere and do not have a corresponding doctoral degree. I have not submitted the doctoral thesis, or parts of it, to another academic institution and the thesis has not been accepted or rejected. I declare that I have acknowledged the Doctoral Degree Regulations which underlie the procedure of the Faculty of Mathematics and Natural Sciences of Humboldt-Universität zu Berlin, as amended on 05th May 2015. Furthermore, I declare that no collaboration with commercial doctoral degree supervisors took place, and that the principles of Humboldt-Universität zu Berlin for ensuring good academic practice were abided by.

29th of May 2019, Berlin, Germany **Patricia Silva**

CONTRIBUTIONS

- P Silva[1], M Neumann[1,2,3] and CD Baldus[1,2,3] designed the study and interpreted the data;
- J Ortiz-Tanchez[1] and K Isaakidis[1] did some of the sample processing and DNA extractions;
- M Neumann[1,2,3] and J Hecht[11,12] designed NGS gene panel;
- P Silva[1] and C Schlee[1] prepared NGS libraries;
- A Graf[7], S Krebs[7], H Blum[7] and PA Greif[2,3,4] performed sequencing;
- M Neumann[1,2,3], S Vosberg[2,3,4] and PA Greif[2,3,4] pre-processed sequencing data;
- MP Schroeder[1] pre-processed Illumina BeadChip arrays;
- P Silva[1] and MP Schroeder[1] run DMR algorithm and its statistical analysis;
- P Silva[1] examined pre-processed data and performed final processing on NGS sequencing and Illumina BeadChip array data;
- P Silva[1] designed and performed statistical analyses for all processed data;
- P Silva[1], K Isaakidis[1] and T Hartung[1] confirmed mutations;
- S Türkmen[5,6] and C Thiede[2,3,9] were involved in clinical diagnostics;
- G Ehninger[2,3,9], C Röllig[2,3,9], C Müller-Tidow[8], H Serve[2,3,10], WE Berdel[13], M Neumann[1,2,3], LR Fransecky[1] and CD Baldus[1,2,3] were involved in patient care and provided clinical and follow-up data;
- G Ehninger[2,3,9], C Röllig[2,3,9], C Müller-Tidow[8], H Serve[2,3,10] and WE Berdel[13] are primary investigators of the SAL registry;
- P Silva[1] implemented outcome analyses of the clinical data;
- P Silva[1] and CD Baldus[1,2,3] wrote the manuscript in Leukemia and the abstract in Blood Journal.

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ABSTRACT [English]

Despite advances in the characterization of molecular alterations in younger acute myeloid leukemia (AML) patients, comprehensive studies in elderly AML are lacking. Thus, we investigated genetic and epigenetic alterations and probed for specific signatures to understand the unfavorable outcomes of elderly AML. We studied 93 AML patients (65 to 90 years old), enrolled in the Study Alliance Leukemia (SAL) registry (SAL elderly AML). To capture a broad spectrum of alterations, we sequenced 555 genes on an Illumina HiSeq2000 platform and investigated DNA methylation profiles using the Illumina 450K array.

Overall, we detected 814 molecular alterations in 281 genes, with a median of 7 genes mutated per patient. Particularly high mutation frequencies were identified for *DNMT3A* (33%), *TET2* (24%), *SRSF2* (23%) and *ASXL1* (21%). We observed frequent alterations in epigenetic regulators (85%) and in splicing factors (38%). Notably, SAL elderly AML patients with mutations in *DNMT3A* or DNA repair genes (in absence of mutations in *NPM1* or splicing factors) had an inferior survival of only 9 months (compared to 17 months for the remaining patients).

In addition, for the analysis of elderly AML DNA methylation, we integrated the SAL cohort with TCGA methylation data for comparisons of methylation levels to younger patients. A distinct DNA methylation profile was observed in older AML patients, which correlated with the presence of mutations in *IDH1/2*, *RUNX1* and *ASXL1* and epigenetic similarities with TP53/Complex samples. The differential methylated regions of elderly AML (compared to younger AML samples) were shown to overlap genes from several pathways that are hallmarks of both age and cancer. Subdivisions of elderly AML showed that *IDH1/2* or *DNMT3A/NPM1/FLT3* mutated samples had diverging epigenetic signatures.

In conclusion, we unraveled distinct patterns of genetic alterations and correlated specific epigenetic profiles of elderly AML to high rate mutated epigenetic regulators. This molecular categorization underscored the distinct biology and the need for specific therapeutic approaches in elderly AML.

ABSTRACT [German]

Die molekulare Charakterisierung von genetischen Veränderungen der Akuten Myeloischen Leukämie (AML) wurde vor allem bei jungen Patienten in den letzten Jahren vorangebracht, hingegen fehlen eingehende Analysen für die AML bei älteren Patienten. Entsprechend ergab sich hieraus die Rationale, genetische und epigenetische Alterationen bei älteren AML Patienten zu untersuchen, um spezifische Signaturen zu identifizieren, die möglicherweise die schlechteren Behandlungsergebnisse bei älteren AML Patienten erklären. Hierzu untersuchten wir 93 AML Patienten (Alter: 65 bis 90 Jahre) aus dem Study Alliance Leukemia (SAL) Register (SAL elderly AML). Um ein breites Spektrum von Alterationen abzudecken, wurden 555 Gene auf der Illumina HiSeq2000 Plattform sequenziert und DNA Methylierungsprofile mittels dem Illumina 450K Array untersucht.

Insgesamt wurden 814 molekulare Alterationen in 281 Genen detektiert (im Median 7 mutierte Gene pro Patient). Besonders hohe Mutationsfrequenzen wurden in den Genen *DNMT3A* (33%), *TET2* (24%), *SRSF2* (23%) und *ASXL1* (21%) notiert. Alterationen in epigenetischen Regulatoren (85%) und in Genen, die in Splicing involviert sind (38%), wurden gehäuft beobachtet. Beachtenswerter Weise wiesen ältere AML Patienten mit Mutationen in DNMT3A oder DNA Reparaturgenen ein geringere Lebenserwartung von nur 9 Monaten auf (im Vergleich zu 17 Monaten in der restlichen Kohorte).

Darüber hinaus integrierten wir die Meythlierungsdaten aus der SAL Kohorte mit denen der TCGA Kohorte, umso die Methylierungsmuster von älteren mit denen von jüngeren Patienten zu vergleichen. Es konnte ein distinktes Methylierungsprofil in den DNA Proben von älteren AML Patienten nachgewiesen werden, welches mit Mutationen in *IDH1/2*, *RUNX1* und *ASXL1* korrelierte. Die im Vergleich zu jüngeren AML Patienten unterschiedlich methylierten Regionen bei älteren AML Patienten überlappten mit Genen verschiedener Signalwege, die Hallmarks von Alterungsprozessen und Krebs entsprechen. Weitere Analysen zeigten zudem divergente epigenetische Signaturen zwischen Proben mit *IDH1/2* Mutationen und *DNMT3A*/*NPM1*/*FLT3* mutierten Proben bei älteren AML Patienten.

Zusammenfassend konnten wir für die Kohorte älterer AML Patienten distinkte genetische Alterationen nachweisen und mit spezifischen Profilen von Mutationen in epigenetischen Regulatoren korrelieren. Diese molekulare Kategorisierung unterstreicht distinkte biologische Mechanismen in älteren AML Patienten und die

Notwendigkeit von spezifischen Therapieansätzen für diese Kohorte mit ungünstiger Prognose.

KEY POINTS

- Elderly AML is a distinct entity with a high frequency of mutations in spliceosome components, epigenetic regulators and DNA repair factors.
- Enrichment of mutations in the DNA repair model postulates treatment resistance and poor outcome in about ¼ of patients.
- Elderly AML has aberrant DNA methylation profiles associated with molecular alterations in *IDH1/2*, *ASXL1* and *RUNX1*.
- The epigenetic patterns of elderly AML related these disease to methylation patterns of poor prognosis involving changes in genes known to be involved in age and cancer/AML processes.
- High frequency of mutations in *IDH1/2* and *DNMT3A* observed in the SAL elderly AML translated into opposite patterns of methylation and drive distinct clusters.
- Elderly AML is in need of different therapeutic approaches to address its molecular specificities and resistance mechanisms.

TERMS AND ABBREVIATIONS

Abbreviation	Full term
2-HG	D-2-hydroxyglutarate
AKT	AKT Serine/Threonine kinase
ALL	Acute lymphoblastic leukemia
alloSCT	Allogenic stem cell transplantation
AML	Acute myeloid leukemia
APL	Acute promyelocytic leukemia
Ara-C	Cytarabine
ATM	ATM Serine/Threonine kinase
ATR	ATR Serine/Threonine kinase
ATRA	All-Trans-Retinoic acid
Aza	Azacitidine
b	Bases, unit (kb=Kilo bases, Mb=Mega base)
biCEBPA	Biallelic mutated CEBPA
BM	Bone marrow
bp	Base pair, unit
BSC	Best supportive care
CBF	Core-binding factor
CDC42	Cell Division Cycle 42
CHIP	Clonal hematopoiesis of indeterminate potential
CHK	Checkpoint kinase
chr	Chromosome
CLL	Chronic lymphoblastic leukemia
CML	Chronic myeloid leukemia
CN-AML	Cytogenetically normal AML
CNS	Central nervous system
COSMIC	Catalogue of somatic mutations in cancer
CpGs	Cytosine-phosphate-guanine sequential nucleotides
CTCF	CCCTC-Binding Factor
DMRs	Differentially methylated regions
DNA-PK	DNA dependent protein kinase catalytic subunit, signaling pathway
DOT1L	DOT1 Like histone lysine methyltransferase
ECOG	Eastern Cooperative Oncology Group
EFS	Event-free survival
ELN	European LeukemiaNet
EMA	European Medicines Agency
ERRBS	Enhanced reduced representation bisulfite sequencing
FAB	French-American-British
FDA	Food and drugs administration
FDR	False discovery ratio
FGFR	Fibroblast growth factor receptor
FI	Functional interaction
FISH	Fluorescence in situ hybridization
GO	Gene Ontology

Gene Symbol	Gene name
ABL1	ABL Proto-Oncogene 1, Non-Receptor Tyrosine Kinase
ACTG1	Actin Gamma 1
APC	APC, WNT Signaling Pathway Regulator
APOB	Apolipoprotein B
ARID1A	AT-Rich Interaction Domain 1A
ASXL1	Additional sex combs-like 1
ATG16L1	Autophagy Related 16 Like 1
ATG5/7	Autophagy Related 5 or 7
ATM	Ataxia telangiectasia mutated
ATR	Ataxia telangiectasia and Rad3 related
BAIAP2	BAI1 Associated Protein 2
BCOR	BCL6 Corepressor
BCORL1	BCL6 Corepressor Like 1
BCR	BCR, RhoGEF And GTPase Activating Protein
BRCA2	Breast cancer 2
C7orf13	Long Intergenic Non-Protein Coding RNA 1006
CBFB	Core-Binding Factor Subunit Beta
CBL	Cbl Proto-Oncogene
CD34	Hematopoietic Progenitor Cell Antigen CD34
CEBPA	CCAAT/enhancer-binding protein-alpha
CHK	Checkpoint Kinase
CHMP6	Charged Multivesicular Body Protein 6
CLU	Clusterin
***CREBBP*[*KAT3A*]**	CREB binding protein
DAB2IP	DAB2 Interacting Protein
DDX5	DEAD-Box Helicase 5
DEK	DEK Proto-Oncogene
DNA-PK	DNA-dependent protein kinase
DNMT3A	DNA Methyltransferase 3 Alpha
DSC2	Desmocollin 2
EP300	E1A Binding Protein P300
ERCC2	ERCC Excision Repair 2, TFIIH Core Complex Helicase Subunit
ESR1	Estrogen Receptor 1
EZH2	Enhancer of Zeste 2
FAM127A	Retrotransposon Gag Like 8C
FAM127B	Retrotransposon Gag Like 8A
FAM92A1	Family With Sequence Similarity 92 Member A
FANCC	FA Complementation Group C
FASN	Fatty Acid Synthase
FAT1/2/3	FAT Atypical Cadherin 1, 2 or 3
FLT3	Fms-Like Tyrosine Kinase 3
FSCN2	Fascin Actin-Bundling Protein 2, Retinal
GATA2	GATA Binding Protein 2

HDAC	Histone deacetylase complex
HELP	HpaII tiny fragment enrichment by ligation-mediated PCR
HOXL	HOX-Like subclass homeobox genes
HSCs	Hematopoietic stem cells
HSPCs	Hematopoietic stem and progenitor cells
HyperMRs	Hypermethylated regions
HypoMRs	Hypomethylated regions
INDEL	Insertions and deletions
inv(*)	Inversions, notation
ITD	Internal tandem duplication
JAK-STAT	Janus kinase/signal transducers and activators of transcription
KI	Knockin, of a gene
LDH	Lactate dehydrogenase
LOH	Loss of heterozygosity
LSC	Leukemic stem cell
MAF	Minor allele frequency
MDS	Myelodysplastic syndromes
mon	months
MPNs	Myeloproliferative neoplasms
mTORC1	Mammalian target of rapamycin complex 1
NGS	Next generation sequencing
NKL	NK-like homeobox genes
no.	numbers
NOTCH	Notch, for signaling pathway
NPM1c	c-terminal mutation of Nucleophosmin
OS	overall survival
PARP	Poly(ADP-ribose) polymerase
PB	Peripheral blood
PCA	Principal component analysis
PCR	Polymerase chain reaction
PI3K	Phosphatidylinositol-4,5-bisphosphate 3-kinase
RAS	RAS family, signaling pathway
SAL	Study Alliance Leukemia
sAML	Secondary AML
SEER	Surveillance, Epidemiology, and End Results
SNP	Single-nucleotide polymorphism
SNV	Single-nucleotide variation
Src	Proto-oncogene tyrosine-protein kinase Src
t(*;*)	Translocations, notation
t-AML	Therapy-related AML
TCGA	The Cancer Genome Atlas
TKD	Tyrosine kinase domain
TNF	Tumor necrosis factor
TSG	Tumor suppressor genes

GNAS	GNAS Complex Locus
HIVEP3	Human Immunodeficiency Virus Type I Enhancer Binding Protein 3
HOXB3/4/5	Homeobox B3, B4 or B5
IDH1/2	Isocitrate Dehydrogenase 1 or 2
IGFBP3	Insulin-Like Growth Factor Binding Protein 3
JAG1	Jagged 1
JAK2	Janus Kinase 2
***KIAA1447*[*BAHCC1*]**	BAH Domain And Coiled-Coil Containing 1
KRAS	KRAS Proto-Oncogene, GTPase
***MECOM*[*EVI1*]**	MDS1 And EVI1 Complex Locus
MEIS1	Meis Homeobox 1
MGMT	O-6-Methylguanine-DNA Methyltransferase
MIR155HG	MIR155 Host Gene
***MLL*[*KMT2A*]**	Mixed-Lineage Leukemia
MLLT3	MLLT3, Super Elongation Complex Subunit
MTOR	Mechanistic Target Of Rapamycin Kinase
MYH11	Myosin Heavy Chain 11
***MYST3*[*KAT6A*]**	Lysine Acetyltransferase 6A
NEXN	Nexilin F-Actin Binding Protein
NHLRC1	NHL Repeat Containing E3 Ubiquitin Protein Ligase 1
NPM1	Nucleophosmin
NRAS	NRAS Proto-Oncogene, GTPase
***NSD1*[*KMT3B*]**	Nuclear Receptor Binding SET Domain Protein 1
NUP214	Nucleoporin 214
PARP1	Poly(ADP-ribose) Polymerase 1
PDE4DIP	Phosphodiesterase 4D Interacting Protein
PHF6	PHD Finger Protein 6
PIK3C2B	Phosphatidylinositol-4-Phosphate 3-Kinase Catalytic Subunit Type 2 Beta
PIK3CA	Phosphatidylinositol-4,5-Bisphosphate 3-Kinase Catalytic Subunit Alpha
PIK3R1	Phosphoinositide-3-Kinase Regulatory Subunit 1
PML	Promyelocytic Leukemia
PPM1D	Protein Phosphatase, Mg^{2+}/Mn^{2+} Dependent 1D
RAD21	RAD21 Cohesin Complex Component
RAD23A	RAD23 Homolog A, Nucleotide Excision Repair Protein
RAD51	RAD51 Recombinase
RAD52	RAD52 Homolog, DNA Repair Protein
RARA	Retinoic Acid Receptor Alpha
RHOC	Ras Homolog Family Member C
***RPTOR*[*RAPTOR*]**	Regulatory Associated Protein Of MTOR Complex 1
RUNX1	Runt Related Transcription Factor 1
RUNX1T1	RUNX1 Translocation Partner 1
SCRN1	Secernin 1
SF3B1	Splicing Factor 3b Subunit 1
SMC1A	Structural Maintenance Of Chromosomes 1A

TSS	Transcription Start Site
VAF	Variant allele frequency
WBC	White blood cells
WHO	World Health Organization
WNT	WNT, for signaling pathway
y	Years
α-KG	α-Ketoglutarate

SMC3	Structural Maintenance Of Chromosomes 3
SOCS2	Suppressor Of Cytokine Signaling 2
SRSF2	Serine And Arginine Rich Splicing Factor 2
TEKT2	Tektin 2
TET2	Ten-eleven-translocated 2
TNF	Tumor Necrosis Factor
TP53/63/73	Tumor Protein 53, 63 or 73
TTC12	Tetratricopeptide Repeat Domain 12
U2AF1	U2 Small Nuclear RNA Auxiliary Factor 1
VWA8	Von Willebrand Factor A Domain Containing 8
WT1	Wilms Tumor 1
XRCC3	X-Ray Repair Cross Complementing 3
ZRSR2	Zinc Finger CCCH-Type, RNA Binding Motif And Serine/Arginine Rich 2

Note: An alternative name for a gene is given inside “[ ]”. A “/” corresponds to a pair of different genes. An “-” separates two genes involved in a fusion. The “*” denotes the possibility of several chromosome parts.

CHAPTER 1. INTRODUCTION

1.1 Hematological diseases

"Only if we understand how hematopoietic stem cells age, we can begin exploring opportunities to prevent, delay, or even reverse aspects of the aging process."[3]

The current indicators show the population of the developing world is expected to continue aging as our lifespan is increasing. Therefore, we need to understand and revert the decline of our tissue regenerative capacities to prevent further deterioration of the quality of life of the elderly. If we cannot prevent the aging-associated tissue attrition the frequency of diseases associated with age will carry on rising. Blood disorders are now counted amongst the classes of diseases markedly increased by age, which was not anticipated since the blood system is constantly renewed with several billion cells being produced every day. However, we continuously find more and more explanations for why hematological diseases are so very age-dependent.

1.1.1 Normal hematopoiesis

Blood cells are continuously regenerated by hematopoietic stem cells (HSCs) residing in the bone marrow (BM) of adult mammals, the process is denominated hematopoiesis. These are rare cells (0.01–0.2% of the total BM mononuclear cells in humans) that seldom divide. When they do divide they undergo asymmetrical divisions assuring one of the daughter cells is a new HSC (self-renewal)[4,5] and so keep the number of HSCs stable.

For the normal functioning of the immune system relative amounts of the blood cells from the lymphoid, erythroid and myeloid lineages need to be produced and kept in balance. Therefore, hematopoiesis has to be very tightly regulated.

HSCs give rise to progenitor cells (HSPCs) that become increasingly lineage-restricted and ultimately differentiate into all lineages of mature blood cells (Figure 1.1.1A).

The hierarchy by which the HSCs became committed to a terminally differentiated blood cell has been under assessment in the latest years since the possibility of analysing mutations, gene expression, proliferation and differentiation at a single-cell level have come to challenge the classical model of hematopoiesis[6,7,8,9].

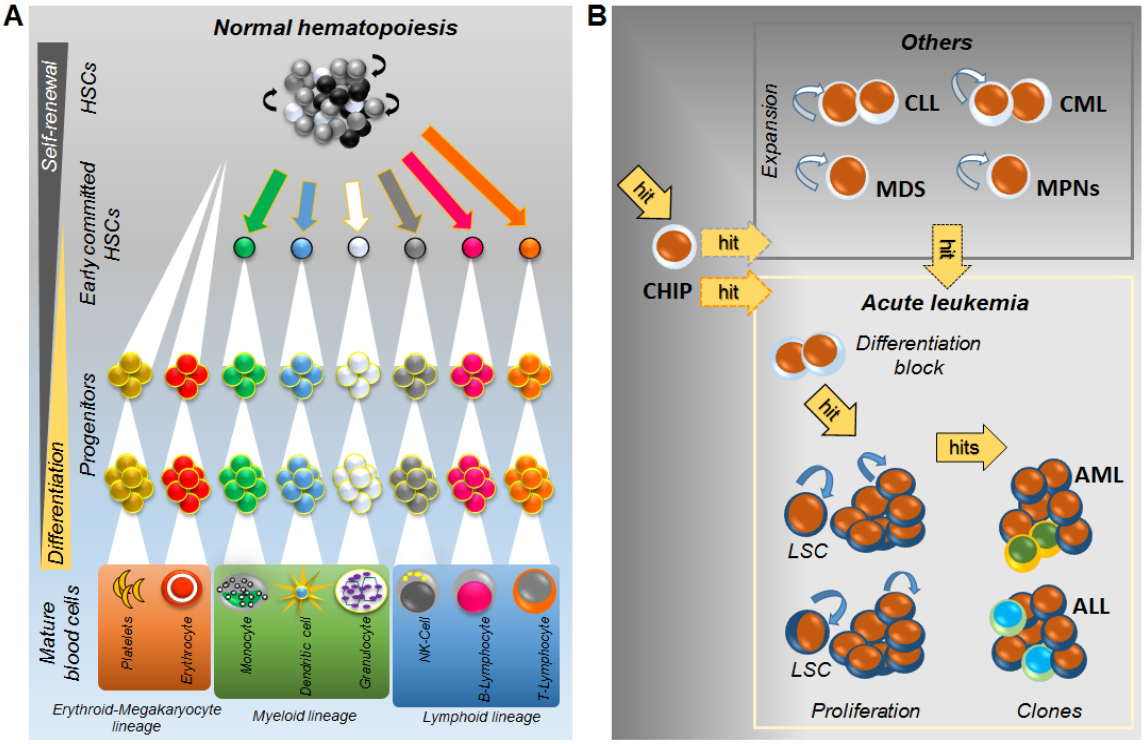


Figure 1.1.1 The normal hematopoiesis and the leukemic stem cell model.
(A) In normal hematopoiesis, rarely dividing hematopoietic stem cells (HSCs) with unlimited self-renewal capacity (indicated by curved arrows) give rise to progenitors (HSPCs). The pool of undifferentiated HSPCs comprises a continuum of transitory cell stages that lack hierarchical structures and discrete lineage-specific progenitor stages. In turn, these intensely proliferative progenitors have progressive states in which self-renewal capacities are decreasing until they ultimately differentiate to all the mature cells of the peripheral blood. (B) The formation of a leukemic stem cell (LSC) in myeloid leukemia may result from mutations in cells in different stages of the hematopoietic hierarchy. Hematologic malignancies are driven by combinations of genetic lesions, the 1st somatic mutation giving rise a pre-leukemic clone: clonal hematopoiesis of indeterminate potential (CHIP), or myelodysplastic syndromes (MDS), or myeloproliferative neoplasms (MPNs). In a pre-leukemic disease phase, cells are genetically unstable, increasing the possibility of further mutations that give rise to the LSCs, developing a chronical or acute disease: acute myeloid leukemia (AML) or acute lymphoblastic leukemia (ALL), chronical myeloid leukemia (CML) or chronic lymphocytic leukemia (CLL). These self-renewing LSCs clonally expand, facilitating the acquisition of additional mutations and development of different leukemic clones. Design by Patricia Silva.

In the classical view, there were bifurcations in commitment steps from multipotent progenitor cells to oligopotent (mixed lineages progenitors) to unipotent (single lineage progenitors). This is currently seen as an early commitment of progenitors with a linear continuum of transitions to the final specification and not requiring several steps, as was previously implied[6,7,9]. Therefore, the existing HSPCs are a heterogeneous pool of cells with long-term durable self-renewal capacities, made of subpopulations of cells already primed for a specific differentiation program[6,8]. This view is being reinforced by the fact the oligopotent progenitors of mixed lineages differentiation potentials were so far not found in the context of hematopoiesis in the

adult bone marrow, but there is evidence they are present in the fetal liver[7]. These new facts delineated a shift in the hierarchy of blood progenitor classes from in utero to adulthood[7]. This readjustment anticipates the existence of high age-plasticity of hematopoiesis.

1.1.2 Hematopoietic disorders

When the hematopoietic process fails and a proliferation of immature bone marrow-derived cells (blasts) occurs that constitutes a hematological disorder (Figure 1.1.1B). These could be pre-leukemic diseases characterized by ineffective hematopoiesis that present phenotypes of cytopenia (lack of certain lineages of mature blood cells).

Nowadays, these are commonly denominated myelodysplastic syndromes (MDS) and myeloproliferative neoplasms (MPNs) and said to evolve from a clonal hematopoiesis of indeterminate potential (CHIP), a newly recognized entity.

The CHIP condition despite possibly being phenotype free (without cytopenia) is defined by the occurrence of molecular alterations in leukemia-associated genes. These are mutations that can be found in the peripheral blood of a small proportion of healthy individuals[10,11]. The pre-leukemic clones in CHIP are cells predisposed to subsequent acquisition of additional genomic alterations. Evolution into an MDS/MPNs is due to further events involving genetic aberrations that do not induce malignant transformations but result in a clonal expansion of mutant progenitors and consequently an increased probability of progression into a leukemic disease.

Therefore, these pre-leukemias can develop into acute leukemia or constitute a chronic form. The most common types of leukemia are: lymphoid leukemias, such as acute lymphoblastic leukemia (ALL) and chronic lymphocytic leukemia (CLL) products of expanded progenitors primed for a lymphoid lineage; or myeloid leukemias, including acute myeloid leukemia (AML) and chronic myeloid leukemia (CML) products of myeloid primed progenitors.

The main model for the development of a full-blown leukemia is the “two-hit hypothesis”[12]. It suggests that the malignant transformation of a progenitor cell to a leukemic stem cell (LSC) that maintains leukemia requires at least two mutations. One of the mutations should guarantee a differentiation blockade and another should ensue uncontrolled proliferation and apoptosis evasion (these are usually revered to as the initiation mutations)[12,13]. It is a simplistic model, it says nothing about the contribution

of epigenetic mechanisms for a fully transformed phenotype and does not have a class for the many mutations found in epigenetic regulators.

In leukemia, it is based mainly on several observations showing that one mutation is usually not enough for leukemic transformations *in vitro* or in mouse models. Recently, the hierarchy of mutations has been studied in several of the leukemic diseases, to identify disease initiation events that are thought to occur early in disease progression. Several early lesions have been identified, for example: DNA methyltransferase 3A (*DNMT3A*) mutations that are found in AML with normal karyotype (CN-AML)[13,14]; *PML-RARA* fusion found in acute promyelocytic leukemia; ten-eleven-translocated 2 (*TET2*) or isocitrate dehydrogenase 2 (*IDH2*)[15] mutations, as well as core-binding factor (CBF) or mixed-lineage leukemia (*MLL*[*KMT2A*])[16] translocations found in 2 other AML types and *ETV6–RUNX1* fusion found in childhood B-ALL[17].

Upon transformation, tumor cells will further acquire multiple genomic or chromosomal aberrations (cooperating mutations)[13], due to their genomic instability. The environmental pressures[18], a tumor is subjected to, result in the natural selection of some clones in detriment of others. Therefore, leukemias are dynamic systems constantly subjected to Darwinian evolution during malignant progression. In leukemia, genomic analyses at the single-cell level are describing clonal compositions, showing which multiple coexisting clones are present at the time of diagnosis[19,20].

Many previous studies had already distinguished the first mutations from the latest clones to arise. These used cytogenetic, molecular and sequencing data obtained from bulk material by estimating variant allele frequencies (VAFs) of the events. Now studies using single-cell genomic analyses are inferring the order of lesions and they found good correlations to the data acquired from the bulks[20,21].

Generally, in AML samples at the time of diagnosis, several initiating mutations have been found in a major clone (that are called driver mutations) but also several later mutations that are not in all the blasts (which were termed passenger mutations). Unlike solid cancers the number of these mutations in hematological disorders looks to be limited, with some recognizable patterns, these observations are giving the field hope to advance treatment options using drugs to target cells with these specific mutations.

1.1.3 Aging of hematopoietic stem cells

Since the HSCs are needed for a lifelong generation of mature blood cells, the adult small population of HSCs needs to be properly maintained during aging for a functional hematopoiesis. The normal HSCs reside in a specialized microenvironment in the bone marrow (called niche) surrounded by supporting stromal cells that provide microenvironmental factors for protection and tight regulation over HSCs survival and function. To guard against internal stresses related to divisions (like telomere erosion) HSCs are in a notorious quiescent state, it has been shown the most primitive of all HSC may undergo only 4 to 5 divisions in the lifetime of a mouse[22]. The burden of proliferation being left for more committed progenitors, HSPCs.

Notwithstanding all guards, human hematopoiesis has shown several phenotypes of age, with older HSCs increasing in frequency, becoming less quiescent[5], exhibiting myeloid-biased differentiation potential compared with young HSC and mobilizing away from the niche[4]. On the molecular level, mouse aged HSCs were observed to lose polarity of proteins like Cdc42, Scribble and Tubulin (in the cytoplasm) and H4K16ac (in the nucleus)[23], whereas human aged HSCs show increased expression of genes involved in myeloid differentiation and lower expression of genes responsible for lymphopoiesis[5].

The notion of continued self-renewal capacities of HSCs had been notoriously hard to reconcile with the aging process. HSCs being tissue-specific stem cells have restricted self-renewal potential and suffer a functional decline with age, what is usually termed of stem cell exhaustion[3,24]. The fact was revealed several times in the hematopoietic system, as transplantation studies showed that HCSs isolated from younger donors have superior powers in repopulating ablated bone marrows[25]. It is believed that for each division an HSC loses some of its stem cell potential and the pool of progenitors with reduced potential increases to compensate[3].

The mechanisms underlying dysfunction of aging HSCs could be cell-intrinsic and cell-extrinsic mechanisms. Most of the phenotypes of HSC associated with age have been attributed to cell-intrinsic mechanisms, although cell-intrinsic changes may also be due to changes that occur in the bone marrow microenvironment. Especially since its shown that malignant myeloid progenitor cells are supported by an altered microenvironment with mesenchymal stromal cells displaying disturbed signaling pathways[26] that could contribute to disease development[27] (including adhesion molecules and metabolic pathways, as well as endocytosis).

However, the most immediate possibility is the occurrence of molecular alterations in the aging HSCs, leading to clonal expansions of dysfunctional progenitors in the human HSPCs pool. The progenitor's contribution to the blood lineages is then skewed to myeloid progeny[5]. The conjecture is that the aged hematopoietic system has clonal hematopoiesis (CHIP) and might then be predisposed to develop myeloid malignancies as a result.

Our understanding of these clonal events took a forward leap in 2012 and 2014 when several reports proved that clonal hematopoiesis is a frequent event in healthy elderly people[10,11,28,29].

In one these studies from a population of 17,182 persons 10% of the individuals older than 70 years of age carried somatic mutations, the most common mutated genes being *DNMT3A*, *TET2*, and *ASXL1*[10]. This was reproduced in another study where more than 2% of the 2,728 individuals had mutations in the blood[11], most of the mutations being associated with advanced age. The vast majority of those mutations (83%) occurred in 19 genes associated with leukemia and lymphoma, nine were recurrently mutated (*DNMT3A*, *TET2*, *JAK2*, *ASXL1*, *TP53*, *GNAS*, *PPM1D*, *BCORL1,* and *SF3B1*)[11].

In both cases, these accumulations do not appear to be random because they were found in specific loci known to be involved in hematological diseases, which could discourage the view of the accumulation of this mutations in the HSPCs being due to DNA damage. Still, the increased mutation loads must indicate persistent DNA damage in these aging cells upon suffering repeated insults.

These studies were transforming. The field became aware that age-related clonal hematopoiesis is a common condition, associated with an increased risk of developing hematologic cancer and increased risk of cardiovascular disease[10,11,29].

This lead to the use of the term “pre-leukemic mutation” for these mutations. They are to be considered part of the evolution to leukemia in patients that suffered from CHIP. A very important fact if we are to understand the specificity of elderly AML since these are the patients that likely suffered from CHIP.

1.2 Acute Myeloid Leukemia

> *"Acute myeloid leukemia (AML) is the most common form of acute leukemia among adults, and it accounts for the largest number of annual deaths from leukemias in the United States. An estimated 21,380 people will be diagnosed with and 10,590 patients will die of AML in 2017."*[30]

This statement was based in the last cancer statistics of 2017 published every year by the American Cancer Society using the Surveillance, Epidemiology, and End Results program (SEER) as a source for long-term population-based incidence data[31]. It reflects the importance of AML within the blood cancers and the need for treatment options in leukemia, as estimations still predict about 50% of patients with AML will die of the disease.

AML is a hematologic malignancy resulting from an oncogenic transformation and blocked differentiation of cells in a myeloid lineage. These immature cells proliferate in the bone marrow, peripheral blood and other tissues. The major cause of the disease symptoms are these accumulations of blasts in the bone marrow for they result in the impairment of normal hematopoiesis. As a result, the anemia causes fatigue and shortness of breath, the neutropenia/leukopenia causes increased susceptibility to infections and the thrombocytopenia (lack of platelets) causes bruises and bleedings. In turn, the invasion of the tissues by blasts can cause specific symptoms depending on the affected organ (like headaches, slurred speech and confusion, belly swelling). The most pressing ones are caused by lack of oxygen in the tissues because blasts clog vessels, making it difficult for erythrocytes to reach them (leukostasis).

Diagnosis of the acute form of a blood disorder traditionally required the presence of 30% of blast cells in the bone marrow[32]. This is now a more relaxed criterion, with the arbitrary number of 20% blasts proposed to be enough for diagnosis of AML, if certain morphologic and cytogenetic features are present in the blood smears or peripheral blood and prior treatment has been excluded[33]. Current recommendations suggest this is possible when some known cytogenetic abnormalities are detected[33]. Namely, if in presence of a number of balanced chromosomal rearrangements and their resulting chimeric fusion genes, which are designated as "recurrent genetic abnormalities", for example t(15;17), t(8;21), inv(16),

or t(16;16) or in a complex karyotype (with 3 or more abnormalities). Reflecting that these are genetic alterations that are recognized to be pathogenic in AML.

Classically, the classification of AML was based more on the morphology of the leukemic blasts and associated dysplasia, surface and intracellular protein expression, and specific chromosomal abnormalities[34]. Nowadays, we know AML as a broad entity and its pathogenesis is highly heterogeneous, involving several combinations of cytogenetic abnormalities, genetic mutations and epigenetic anomalies that were identified over the years (Figure 1.2.1).

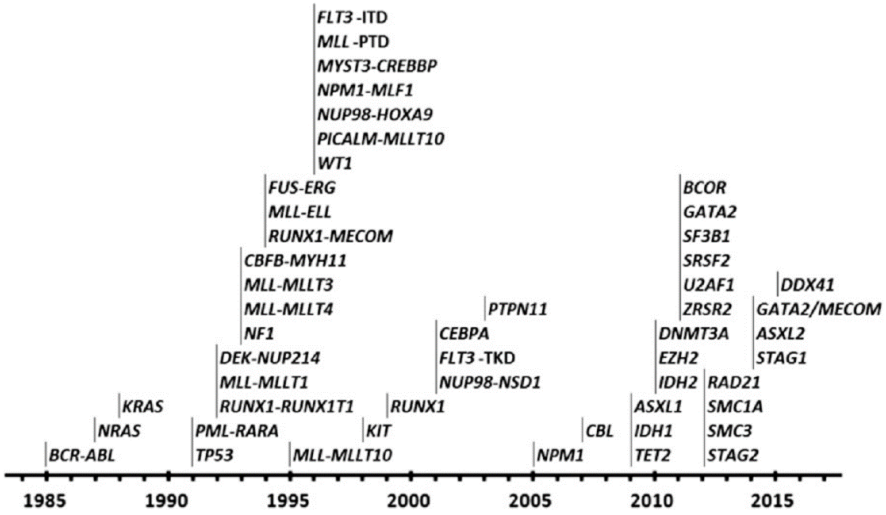


Figure 1.2.1 Progress in defining the molecular landscape of AML.
Timeline of the identification of leukemic fusion genes and mutations underlying the pathogenesis of AML. Adapted from Grimwade *et al.* 2016[35].

As the knowledge of the molecular pathogenesis of AML advanced, several classifications dividing AML entities by cytogenetic aberrations and different diagnostic/prognostic molecular markers have been established[36]. Cytogenetic and molecular cytogenetic studies have long ago found recurrent balanced and unbalanced translocations and inversions that constitute separate molecular classes in AML and were associated with distinct outcomes[36]. However, up to now nearly 50% of AML samples still present a normal karyotype and many of these genomes lack structural abnormalities, even when assessed with high-density comparative genomic hybridization or single-nucleotide polymorphism (SNP) arrays[37]. Therefore, several studies started using target sequencing and found recurrent somatic mutations. Currently, the emerging high throughput sequencing techniques are providing extensive molecular data on AML. These recent updates are characterizing novel AML subgroups based on patterns of somatic mutations[1,37,38,39].

The leukemic field expects that grouping AML into genetically defined subtypes could provide insights into new treatment designs or at least outcome predictions. The big amount of research effort being applied to that objective in recent years has reflected the importance of this goal. In the duration of our study, several other studies have tried to get at this objective using large cohorts of AML to categorize AML patients[34,37,38,40].

1.2.1 AML diagnosis

The diagnosis of AML requires a trained hematologist for the analysis of the morphology of the cells present in blood and marrow smears. They are required to count 200 leukocytes in a blood smear and 500 nucleated cells in marrow aspirate smears[36] and report the percentage of cells with blast morphology found.

Apart from these, specific lineage markers are used to help define an immunophenotype for the disease. So, flow cytometric assessments of the expression of cell-surface and cytoplasmic markers are important. These markers should establish the amounts of myeloid progenitors (using CD34, CD117, CD33, CD13, and HLA-DR) and of several lineages: granulocytic (using CD65 and cytoplasmic myeloperoxidase), monocytic (using CD14, CD36 and CD64), megakaryocytic (using CD41and CD61) and erythroid (using CD235a and CD36)[36].

Furthermore, for a suspected AML, the diagnostic workup has to include conventional cytogenetic tests[36]. Many patients with AML will have cytogenetic aberrations which can be detected through karyotyping or fluorescent in situ hybridization (FISH). Therefore rapid testing of common rearrangements should be done for they are useful for the recommendation of suitable therapies, like *PML-RARA*, *CBFB-MYH11*, *RUNX1-RUNX1T1*, *BCR-ABL1*. Additionally, rearrangements involving genes encoding epigenetic regulators (*MLL*[*KMT2A*], *CREBBP*[*KAT3A*] and *NSD1*[*KMT3B*]) or nuclear pore complex components (*NUP214*, *NUP98*) although less frequent can be found.

Recently, molecular testing has also entered clinical practice, mostly using reverse transcriptase–polymerase chain reaction (RT-PCR) for the detection of some of the recurrent rearrangements (*RUNX1-RUNX1T1*, *CBFB-MYH11*, *MLLT3-MLL*, *DEK-NUP214*) but also some mutations with prognosis predictions, like: alterations of nucleophosmin (*NPM1*) that change tryptophan residues in the protein C-terminus (*NPM1c*), biallelic mutations of CCAAT/enhancer-binding protein-alpha (*biCEBPA*) and Fms-Like Tyrosine Kinase 3 (*FLT3*) internal tandem duplications (*FLT3*-ITD).

The most recent recommendations indicate that confirmation of AML should further include tests of mutations in the tyrosine kinase domain of FLT3 (*FLT3*-TKD, codons D835 and I836), *RUNX1*, *TP53* and *ASXL1*[36]. Sequencing all these genes will be more feasible by applying gene panel next generation sequencing (NGS) techniques and will soon become the norm, due to the decreasing costs of the NGS.

1.2.2 AML classifications

The first classifications of leukemia proposed were based in morphological features, for instance, the French-American-British (FAB) co-operative group[32] classification, but their power for clinical outcome prediction was very limited. The identification of leukemia-associated chromosomal translocations and inversions paved the way to the World Health Organization (WHO) classification of myeloid neoplasms and acute leukemia in 2001[41] and for its revision in 2008[42]. These classifications of AML were mostly cytogenetic based. They were used to guide the care of patients with AML for many years, for they allowed the division of patients into groups with favorable, intermediates, or adverse prognosis.

Since then, the advances in NGS provided molecular knowledge that led to the WHO updated classification[33] in 2016.

This publication greatly illustrated how, despite the increased knowledge, it remains a struggle to describe the morphological heterogeneity of the AML disease and the AML related diseases. Therefore, the disease was divided into many groups of general morphologic characteristics and these had to be further divided into subgroups with more specific phenotypes and genetic alterations (Table 1.2.1).

Table 1.2.1 WHO 2016 classifications of AML

AML and related precursor neoplasms and acute leukemia of ambiguous lineage classified as by WHO in 2016. Adapted simplification from Arber *et al.*[33] and Döhner *et al.*[36].

AML and related neoplasms
AML with recurrent genetic abnormalities ○ AML with t(8;21)(q22;q22.1); *RUNX1-RUNX1T1* ○ AML with inv(16)(p13.1q22) or t(16;16)(p13.1;q22); *CBFB-MYH11* ○ Acute promyelocytic leukemia with *PML-RARA* ○ AML with t(9;11)(p21.3;q23.3); *MLLT3-MLL*[*KMT2A*] ○ AML with t(6;9)(p23;q34.1); *DEK-NUP214* ○ AML with inv(3)(q21.3q26.2) or t(3;3)(q21.3;q26.2); *GATA2*, *MECOM*[*EVI1*] ○ AML (megakaryoblastic) with t(1;22)(p13.3;q13.3); *RBM15-MKL1* ○ Provisional entity: AML with *BCR-ABL1* ○ AML with mutated *NPM1* ○ AML with biallelic mutations of *CEBPA* ○ Provisional entity: AML with mutated *RUNX1*
AML with myelodysplasia-related changes
Therapy-related myeloid neoplasms
AML, NOS ○ AML with minimal differentiation ○ AML without maturation ○ AML with maturation ○ Acute myelomonocytic leukemia ○ Acute monoblastic/monocytic leukemia ○ Pure erythroid leukemia ○ Acute megakaryoblastic leukemia ○ Acute basophilic leukemia ○ Acute panmyelosis with myelofibrosis
Myeloid sarcoma
Myeloid proliferations related to Down syndrome ○ Transient abnormal myelopoiesis ○ Myeloid leukemia associated with Down syndrome
Blastic plasmacytoid dendritic cell neoplasm
Acute leukemias of ambiguous lineage
Acute undifferentiated leukemia
MPAL with t(9;22)(q34.1;q11.2); *BCR-ABL1*
MPAL with t(v;11q23.3); *MLL*[*KMT2A*] rearranged
MPAL, B/myeloid, NOS
MPAL, T/myeloid, NOS

1.2.3 AML prognostic factors

Typically, there are several considerations to have when trying to predict a patient resistance to treatment or outcome, mainly because not only the disease factors but also the patient previous and current health state are relevant. Even so, the presence of certain genetic lesions are the most important factors accounting for good or bad outcomes[36].

Foreseeably, research into predictive markers based on the genetic characteristics of the disease has been one of the most important fields of leukemia for many years. However, until now predictive models for live expectancy have only reached 75-80% accuracy[36], with contributions from demographic, clinical and treatment variables being hard to model.

According to current knowledge, prognostic factors have been divided into patient-related factors or AML-related factors[43].

The most important prognostic factor patient-related is increasing age at the time of diagnosis, which has been shown to be an independent predictor of poor outcome[44,45,46] and suggests the effect of unknown age-related factors[47]. Other patient-related factors have been associated to differences in outcomes such as coexisting medical conditions (especially comorbidities) and poor performance status (a measure of patient fitness from the impact of cancer on patients daily living abilities, ECOG score, graded 0-healthy to 5-death). These factors complicate the application of intensive induction chemotherapies and are correlated to increased treatment-related early death[43].

The majority of prognostic factors known are related to the disease itself. One of these factors is white blood cells count (WBC), which when higher than $100x10^9$/L is considered hyperleukocytosis and connected to increased mortality during chemotherapy.

Others are splenomegaly and elevated serum lactate dehydrogenase (LDH), although these appear to provide poor prognosis only in certain cohorts[43].

Furthermore, the AML diagnosed according to the disease history has consistently proved to be of prognostic value. AML can be classified into 3 distinct categories based on clinical ontogeny: secondary AML (sAML) represents the transformation of an antecedent diagnosis of MDS/MPNs; therapy-related AML (t-AML) that develops as a complication in patients with prior exposure to cytotoxic therapies; and *de novo* AML that refers to the absence of any identified hematological disorder or prior exposure to therapy.

The two classes sAML and t-AML have demonstrated poor prognosis predictions, mostly due to increased resistance to current standard chemotherapy leading to low median overall survival probabilities[43,48]. However, the spectrum of genetic lesions of these two classes is different from the *de novo* AML, having shown genetic aberrations typical of the MDSs (like single nucleotide variations, SNVs, in splicing factors) and low frequencies of cytogenetic characteristics of favorable prognosis (like CBF rearrangements)[49]. Therefore, the sAML and t-AML ontogenies are connected to genetic abnormalities that are powerful prognostic factors and might be the responsible factors that confer these particular poor prognoses[49,50].

Since the presence of specific genetic abnormalities still has the most power to predict survival probabilities, genetically based predictors are the most informative

disease-related factors. So far, the prediction of inferior outcomes in a clinical setting has depended on the evaluation of common cytogenetic lesions and 3 molecular genetic markers (*NPM1c* and *biCEBPA* mutations and *FLT3*-ITD). These were factors included in the European LeukemiaNet (ELN) recommendations of 2010[47] that became clinical practice. The 2010 ELN recommendations were done by a panel of international experts, which endeavored to standardize the reports of genetic abnormalities and its correlations to clinical outcomes. They stratified patients into 4 risk categories according to their molecular genetic data (favorable, intermediate-I, intermediate-II and adverse)[47].

The amount of research data accumulated in the last 5 years was calling for a revision of this system. To start with, the 2 groups intermediate-I and II were impossible to distinguish in older patients, which actually are the major group of patients. Additionally, several studies had shown previously unknown genetic markers associated with poor prognosis. As a result, last year the ELN published their update of the recommendations for the groups based on this new data on treatment outcomes[36].

Therefore, the most significant changes were the merging into 3 risk categories (favorable, intermediate and adverse). Typically, patients with favorable risk cytogenetic profiles present balanced structural rearrangements (fusions *PML-RARA*, *RUNX1-RUNX1T1*, *MYH11-CBFB*), and normal karyotype with *biCEBPA* or mutated *NPM1*. Conversely, patients with unfavorable-risk cytogenetic profiles display imbalanced abnormalities (fusions like *DEK-NUP214* or *BCR-ABL1*, rearrangements of *KMT2A*, *MECOM* or *GATA2*, deletion of 5q, abnormal 3q, monosomy of chromosome 7), or three or more complex abnormalities (complex karyotype). This group now includes the poor outcomes of patients with *RUNX1*, *ASXL1* or *TP53* mutations (originating Table 1.2.2).

Table 1.2.2 ELN 2017 risk stratification by genetics characteristics.

Table with the new ELN risk categories simplified from the reported in Bullinger *et al.*[51].

Risk Category	Genetic Lesion
Favorable	○ t(8;21)(q22;q22.1); *RUNX1-RUNX1T1* ○ inv(16)(p13.1q22) or t(16;16)(p13.1;q22); *CBFB-MYH11* ○ Mutated *NPM1* without *FLT3*-ITD or with *FLT3*-ITD^{low} ○ Biallelic mutated *CEBPA*
Intermediate	○ Mutated *NPM1* and *FLT3*-ITD^{high} ○ Wild-type *NPM1* without *FLT3*-ITD or with *FLT3*-ITD^{low} (if without adverse-risk gene mutations) ○ t(9;11)(p21.3;q23.3); *MLLT3-MLL*[*KMT2A*] (takes precedence over adverse-risk mutations) ○ Cytogenetic abnormalities not classified as favorable or adverse
Adverse	○ t(6;9)(p23;q34.1); *DEK-NUP214* ○ t(v;11q23.3); *MLL*[*KMT2A*] rearranged ○ t(9;22)(q34.1;q11.2); *BCR-ABL1* ○ inv(3)(q21.3q26.2) or t(3;3)(q21.3;q26.2); *GATA2*, *MECOM*[*EVI1*] ○ -5 or del(5q); –7; –17/abn(17p) ○ Complex karyotype$^{\Delta}$, monosomal karyotype* ○ Wild-type *NPM1* and *FLT3*-ITD^{high} ○ Mutated *RUNX1* (if without favorable-risk AML gene mutations) ○ Mutated *ASXL1* (if without favorable-risk AML gene mutations) ○ Mutated *TP53*

FLT3-ITD^{low}: low allelic ratio (<0.5); *FLT3*-ITD^{high}: high allelic ratio (≥0.5); semi quantitative assessment of *FLT3*-ITD allelic ratio (using DNA fragment analysis) is determined as the ratio of the area under the curve "*FLT3*-ITD" divided by the area under the curve "*FLT3*-wild-type"; recent studies indicate that acute myeloid leukemia (AML) with *NPM1* mutation and *FLT3*-ITD low allelic ratio may also have a more favorable prognosis, and patients should not routinely be assigned to allogeneic hematopoietic cell transplantation.
$^{\Delta}$Three or more unrelated chromosome abnormalities in the absence of one of the WHO-designated recurring translocations or inversions (ie, t(8;21), inv(16) or t(16;16), t(9;11), t(v;11)(v;q23.3), t(6;9), inv(3) or t(3;3), AML with *BCR-ABL1*).
*Defined by the presence of one single monosomy (excluding loss of X or Y) in association with at least one additional monosomy or a structural chromosome abnormality (excluding core-binding factor AML).

These risk-groups are expected to change soon, for they are predicated on patients being treated with the conventional chemotherapies. They could soon become obsolete due to the good results of new therapies that are being directed to certain molecular alterations.

1.2.4 AML treatments

Since 1970, when it was recognized the activity of cytarabine (Ara-C) and anthracyclines against blasts, major progress has been made in the treatment of patients with AML[52]. The recommendation is, as soon as the diagnostic workup of a patient has been completed with an AML diagnosis, intensive chemotherapy should start, provided he/she can tolerate it[36]. The standard induction therapy currently used consists of 7 days of cytarabine (100-200 mg/m^2 continuous IV) and 3 days of an anthracycline (daunorubicin, at least 60 mg/m^2, idarubicin, 10-12 mg/m^2, or mitoxantrone, 10-12 mg/m^2), this treatment is commonly denominated "7+3". This regime as yield an average 35 to 40% long-term cures in young patients with AML (meaning patients 60 years old or younger)[43].

The major accomplishments in AML therapy in the recent past are due to advances in the understanding of AML heterogeneity since improvements have been obtained only from matching particular treatments to specific subgroups of AML[52]. For instance, regimens that have resulted in cure rates 80% to 90%, like treatments with All-Trans-Retinoic Acid (ATRA) and arsenic trioxide[52] in acute promyelocytic leukemia (APL), that target its PML-RARA and eliminated the need for chemotherapy[53,54]. The same is happening for treatments using fludarabine, high-dose cytarabine, anthracyclines[55] or gemtuzumab ozogamicin[56,57] in patients with the CBF type of AML[52].

More recently, genetic studies have provided insights into other molecular targets, for example, *FLT3*-ITD that had consistently been associated with poor prognosis mostly due to a high relapse rate[58]. The molecular characterization of this target has led to the development of a novel therapy soon to be integrated into clinical practice. It consists in the addition of Midostaurin (multi-targeted kinase inhibitor) to standard chemotherapy, which has shown to significantly prolong overall and event-free survival (OS and EFS) among patients with *FLT3* mutations[59] and is now approved for treatment of this group of patients.

Other targeted therapeutic approaches have promised improvements in patient outcomes, including epigenetic directed treatment approaches with hypomethylating agents[60,61]. The first of this class to be applied were azacitidine (Aza) and decitabine (5-aza-2-deoxycytidine), licensed by the Food and drugs administration (FDA) for their demethylation activities (to use in MDS)[62] and registered by the European Medicines Agency (EMA; for use in elderly AML patients aged 65 years and older)[63,64]. These agents promote a broad epigenomic reprogramming, which has been shown to reverse the hypermethylations at promoters of tumor suppressor genes (TSG) and allow for normal differentiation. Interest in these therapies for AML has been increasing due to reports that several enzymes with special roles in the epigenetic regulation of gene expression and initiation of chromatin remodeling are affected by somatic mutations in AML. Since the epigenetic alterations in AML are not only responsible for silencing of TSGs, but also for several transcription activations, it has been very difficult to identify which subset of AML may benefit most from such broadly hypomethylating therapies.

More directed therapies are underway, for instances specifically targeting DNA methyltransferases (DNMTs) which one of the protein families affected by mutations. The mammalian DNMTs are DNMT1, DNMT3A and DNMT3B, which together with accessory proteins, like DNMT3L, are responsible for the acquisition of methylation

patterns during gametogenesis, embryogenesis and somatic tissue development. The DNMTs affected in AML are mostly DNMT3A, with the highest frequency of genetic alterations[37], and DNMT3B for which overexpression in samples represents lower survival probability[65]. At least DNMT3A is an essential DNA methylation regulator thought to have a severe impact on DNA methylation patterns[66]. Despite that, specific DNMT3A inhibitors have so far not been applied and combinations of these with the histone deacetylase complex (HDACs) inhibitors are still under evaluation[67].

In contrast, specific inhibitors of mutant isocitrate dehydrogenases (IDHs) were developed much faster[68,69,70]. IDHs are enzymes involved in metabolism, by catalyzing the conversion of isocitrate to α-ketoglutarate (α-KG) in the tricarboxylic acid cycle (TCA cycle), for which point mutations were found in AML (in the genes *IDH1* or *IDH2)*[1,71,72]. The latest developments of IDH inhibitors have led to the approval of Enasidenib (AG-221)[73], by the FDA[74], for the treatment of relapsed and refractory *IDH2* mutated AML patients. For the corresponding *IDH1* mutated group of patients other IDH inhibitors are already in clinical trial. Specifically, Ivosidenib (AG-120) has shown good results inducing remissions in relapsed or refractory AML patients and was a well-tolerated treatment[75].

All the new treatments in AML being developed are expected to increase patients' survival and promise a bright future to the field. A future that was not within sight during the last decades, when progress was only due to improvements in best supportive care and in stem cell transplantation techniques. This is now a foreseeable future due to hard work in the analysis of the mechanisms by which certain genetic and epigenetic aberrations drive AML.

1.3 Genetic groups of AML

> *"The recent quantum leap in sequencing technology, aided by a dramatic reduction in sequencing costs, has allowed for the generation of previously unparalleled amounts of information annotating mutational landscapes and their evolution across multiple cancers. As befits the first cancer genome sequenced, nowhere is this more apparent than in AML."*[35]

Most of the genetic alterations that initiate AML were only discovered after 2008 when the first whole cancer genome sequence and its matching normal tissue was published[76]. The study, done by Ley *et al*.[76], reported the sequencing of a CN-AML sample. They found this patient had non-synonymous somatic mutations that could be relevant for AML pathogenesis and would not have been sequenced otherwise, for they were unforeseen[76]. This demonstrated broad NGS approaches were required if we were to discover new recurrent somatic mutations to identify candidate genes for treatment development.

The first comprehensive NGS study genetically characterized 200 AML patients with a *de novo* AML disease using a whole-exome sequencing approach. It was performed by the consortium The Cancer Genome Atlas[37] (TCGA) in 2013. The study reported, 154 genes had more than 1 non-synonymous somatic mutation and an additional 1,623 genes had a validated coding mutation in one sample. They observed 23 genes to be significantly mutated, including genes that were established as being relevant to AML pathogenesis (e.g., *DNMT3A*, *FLT3*, *NPM1*, *IDH1*, *IDH2*, and *CEBPA*), along with genes that have only recently been implicated in AML pathogenesis (*U2AF1*, *EZH2*, *SMC1A*, and *SMC3*).

The genes frequently mutated in AML were grouped into nine larger sets or pathways (Figure 1.3.1): transcription factor fusions, the *NPM1* gene, tumor suppressor genes, DNA methylation-related genes/epigenetic regulators, signaling genes, chromatin-modifying genes, myeloid transcription factor genes, cohesion complex genes, and spliceosome complex genes/splicing factors[37].

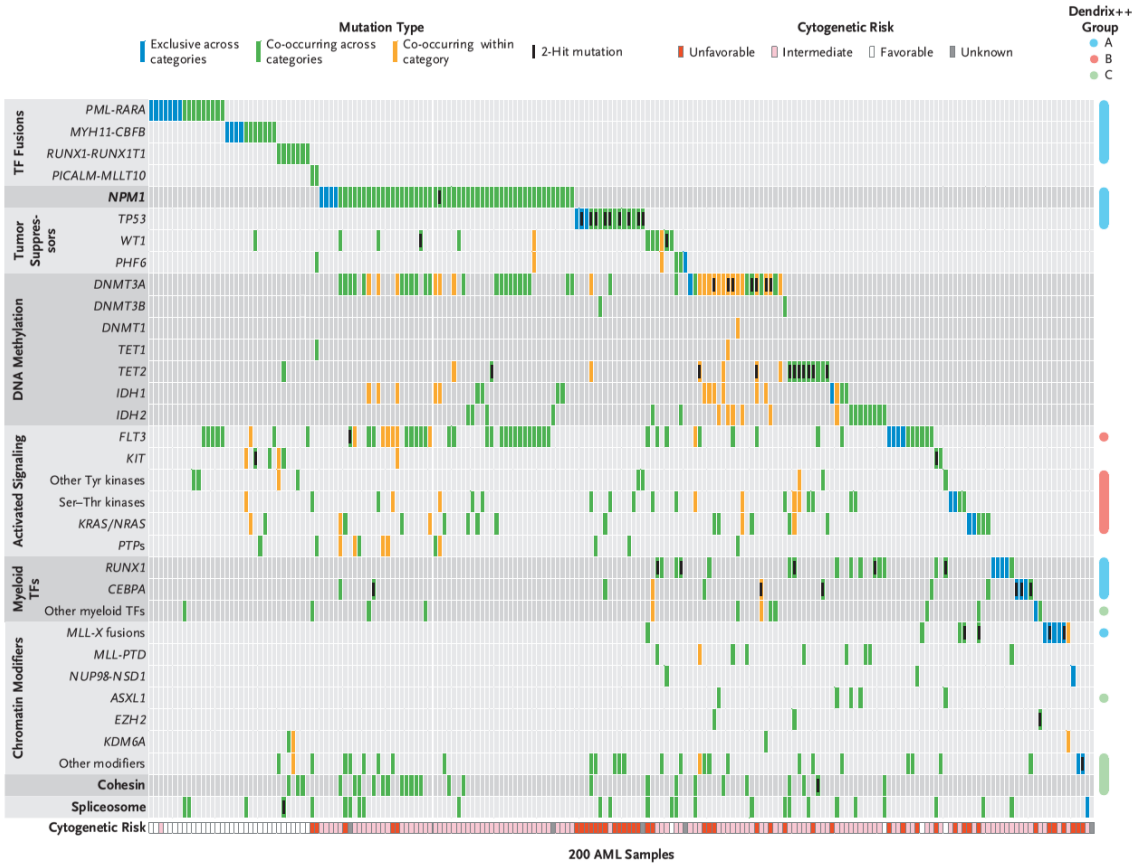


Figure 1.3.1 Organization of mutations into categories of related genes.
"Shown are somatic, nonsynonymous mutations in individual genes and sets of genes, grouped into nine categories, including one single-gene category, as labeled on the left. Of the 200 samples evaluated, 199 (>99%) had at least one mutation in one of the listed genes or sets. Blue boxes indicate mutations that are exclusive across all categories; green boxes, mutations that co-occur in the same sample across different categories; and orange boxes, mutations that co-occur in the same sample in the same category. Computational analysis with the use of the Dendrix++ algorithm identified three significant, mutually exclusive groups of genes, annotated on the right as groups A, B, and C. The cytogenetic risk for each patient is shown at the bottom of the chart. Ser–Thr denotes serine–threonine, TF transcription factor, and Tyr tyrosine." Adapted from TCGA study of 2013[37].

These gene sets were used to examine patterns of mutual exclusivity and co-occurrence of mutations within and between groups. In this analysis, TCGA identified combinations of mutations in samples that occur in non-random patterns. The most prominent relation was the significant co-occurrence between mutations in *FLT3*, *DNMT3A*, and *NPM1*, which they considered to constitute a new AML group[37].

At present, several studies (including ours) have applied target re-sequencing techniques and confirmed several genes to be frequently mutated, with patterns of co-occurrence or mutual exclusivity quite similar to the discovered by TCGA[1,37,39,51].

One of the latest studies investigated 111 genes by NGS studying 1,540 AML patients[39]. They found 76 genes mutated with the large majority of patients having at least 2 driver mutations. The co-mutational patterns they found resulted in a genetic-

based classification of 11 non-overlapping AML classes, some of them newly recognized classes (Figure 1.3.2). These classes were primarily made of the different known fusion genes or the presence of some class defining mutations: *NPM1*; *biCEBPA*; *TP53*/chromosomal aneuploidies; chromatin and RNA-splicing regulators; *IDH2*^{R172} alterations. Other samples were segregated into groups without driver mutations, without class defining mutations or included in a group for the rare fusions[39].

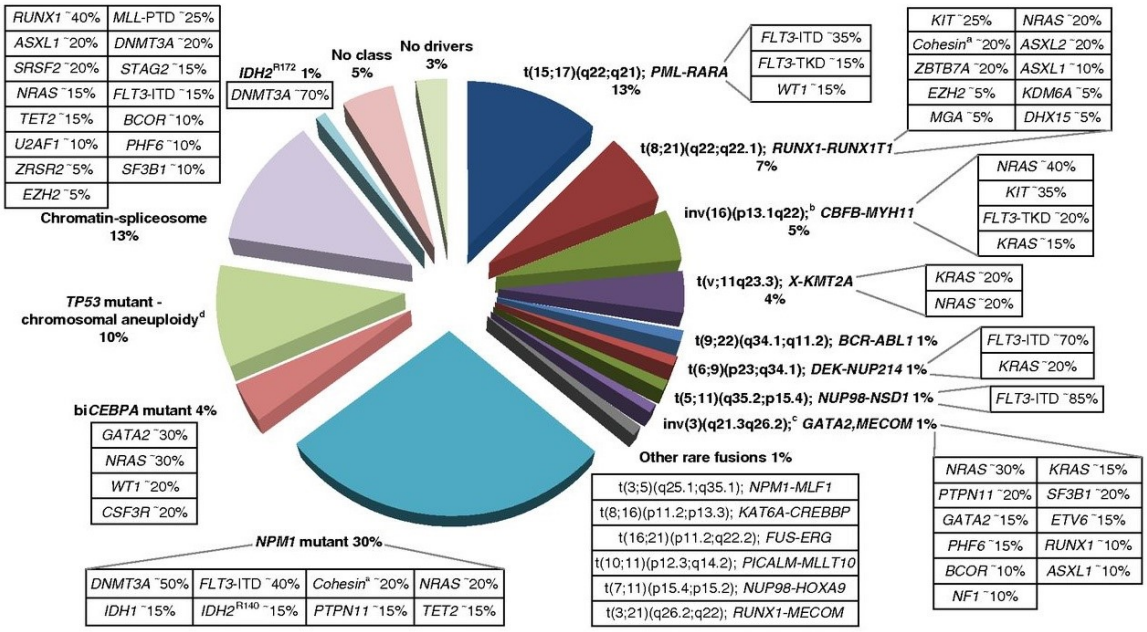


Figure 1.3.2 Molecular classes of AML and concurrent gene mutations in adult patients.

"For each AML class denoted in the pie chart, frequent co-occurring mutations are shown in the respective boxes. Data on the frequency of genetic lesions are compiled from the databases of the British Medical Research Council (MRC), the German-Austrian AML Study Group (AMLSG), and from selected studies. [a], indicates cohesion genes including RAD21 (~10%), SMC1A (~5%), and SMC3 (~5%); [b], inv(16)(p13.1q22) or t(16;16)(p13.1;q22); CBFB-MYH11; [c], inv (3)(q21.3q26.2) or t(3;3)(q21.3;q26.2); GATA2, MECOM(EVI1); and [d], TP53 mutations are found in ~45%, and complex karyotypes in ~70% of this class". The structure of the pie chart is adapted from Grimwade *et al.*[35] generated by Adam Ivey (King's College London, London, United Kingdom) and presented in Döhner *et al.*[36], with the data from Papaemmanuil *et al.*[39].

The statistical power of profiling 1,540 patients gave them the opportunity to attribute different survival probabilities to several of these groups, by separating patients with gene fusions from others[39].

They found class defining gene fusions had different probabilities of survival between them, which was expected. They further report, individual mutational patterns in absence of gene fusions could also have a distinct prognosis and small classes of

groups defined by the presence or absence of drivers can also be discriminated (Figure 1.3.3)[39].

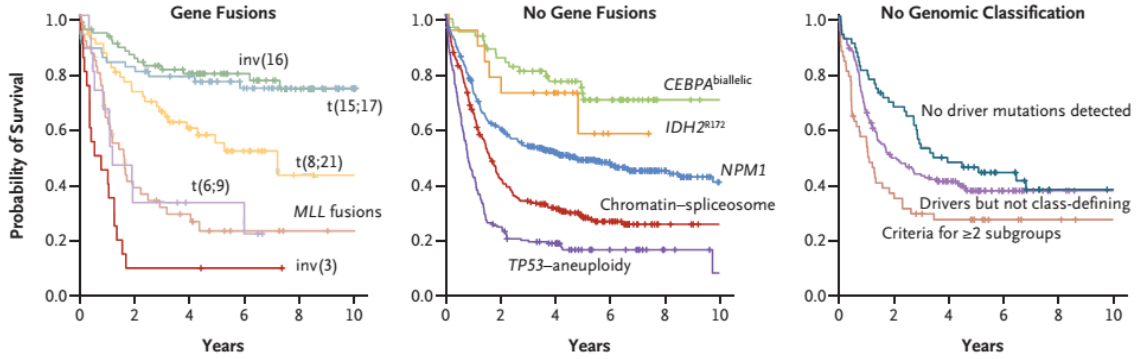


Figure 1.3.3 Different overall survival of molecularly classified groups of AML.
The panels show Kaplan–Meier curves for overall survival among patients in the 11 genomically defined subgroups and patients who did not have a straightforward classification. Adapted from Papaemmanuil *et al.*[39].

All data gathered with the new genetic analysis of AML cohorts brought the field close to understanding the complex molecular patterns of the disease. Since underneath the heterogeneity certain genetic alterations make up specific subsets of AML we can now set to understand what drives the disease in each case. Furthermore, studies can dedicate to mapping tumor dynamic progressions, using its multiple driver mutations and coexisting competing clones that mark disease evolution over time[51].

However, studying the time dimension has still not been enough. Ever since chromosomal translocations and fusions were discovered pathologic in AML, it was suspected that the disease complexity does not end at the genetic level. Many of them affected chromatin modulators as *MLL*[*KMT2A*], *CBP*[*KAT3A*], and *NSD1*[*KMT3B*], but still, that was only a small fraction of AML.

Nowadays, we know that chromatin is affected in most of the AML samples. The whole genome or exome sequencing of AML and the massive parallel sequencing studies led to the identification of mutations in many genes that affect the epigenetic landscape. These modulators of transcription include a broad spectrum of regulators of DNA-methylation or demethylation and modifiers of histone acetylation or methylation (Figure 1.3.4). Recurrent mutations in AML and MDS were found in DNA methyltransferases (*DNMT1/3A*), isocitrate dehydrogenases (*IDH1/IDH2*), methylcytosine dioxygenases of the ten-eleven-translocated family (*TET1/2*), human homologs of the *Drosophila* polycomb complex such as Enhancer of Zeste 2 (*EZH2*) and additional sex-combs like genes (*ASXL1/2*). Moreover, there are a number of examples of epigenome modulators that are not directly mutated but have been

indirectly implicated in AML pathogenesis, like the histone methyltransferase DOT1L that interacts with the MLLTs which in turn are usually MLL-fusion partners.

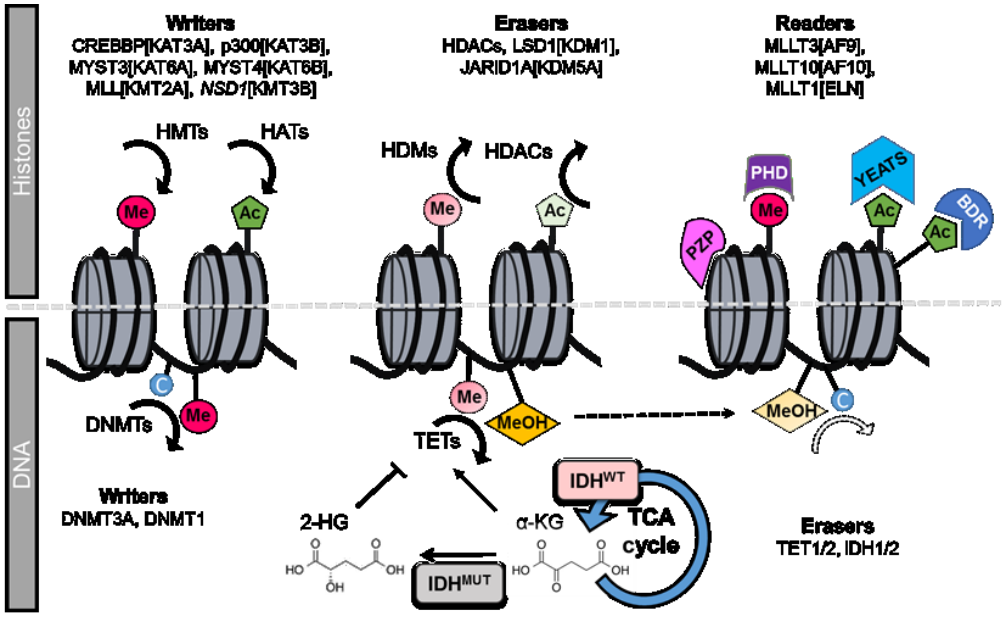


Figure 1.3.4 Types of epigenetic regulators mutated in AML that change or read epigenetic marks in DNA or histones.
Epigenetic "writers" such as histone methyltransferases (HMTs), and histone acetyltransferases (HATs), deposit methylation and/or acetylation on histones, as do DNA methyltransferases on DNA. These epigenetic marks may be removed by epigenetic "erasers," including histone demethylases (HDMs) and histone deacetylase complexes (HDACs). Removal of methylation from DNA is a stepwise process that involves many intervenient enzymes directly and indirectly affected by mutations in AML (TETs and IDHs). Epigenetic "readers" are highly specialized proteins that specifically bind to distinct epigenetic marks to convey this information to downstream effectors. Abbreviations of molecules and domains: alpha-ketoglutarate (α-KG); 2-hydroxyglutarate (2-HG); bromodomains (BRDs); plant homeodomain (PHD); PHD-zinc finger-PHD (PZP). In brackets are alternative gene names/symbols. Design by Patricia Silva.

***DNMT*:** Mutations in *DNMT3A* are observed in 12–22% of AML in most cohorts and usually are heterozygous mutations. Approximately 60% of all *DNMT3A* mutated AML patients harbor a missense mutation in the arginine 822 residue that diminishes its methyltransferase activity while reducing its binding affinity to DNA. These mutations have a dominant negative function over the wild-type DNMT3A protein[77]. The majority of the remaining mutations of *DNM3A* are nonsense or frame-shift mutations in the protein-coding region that led to premature truncations of the protein.

TET: The ten-eleven translocation (TET) family of proteins regulates DNA methylation through the conversion of 5-methylcytosine to 5-hydroxymethylcytosine, which constitutes a step in a complex reaction for removal of DNA methylation[78]. The 5-hydroxymethylcytosine is thought to block the binding of proteins that mediate

transcriptional silencing by recognizing methylated DNA and therefore it present in sites of active transcription. Currently, the higher mutation frequency is in *TET2*, with around 14% up to 22% in some cohorts. Most of these mutations are insertions or deletions, but point-mutations also have been found. *TET2*-mutations constitute loss-of-function leading to decreased 5-hydroxymethylcytosine sites. This results in aberrant hypermethylation and therefore decreased expression of key differentiating enzymes and inhibition of normal cellular differentiation[79,80].

IDH: AML samples were found to have frequent point mutations in *IDH1* Arg132 (R132) and *IDH2* Arg172 or Arg140 (R172 or R140)[1,37,38,39]. *IDH1* and *IDH2* mutations are found at a frequency of about 10% each and more common in CN-AML. It was proposed that these mutations in the *IDH* genes constitute a gain-of-function. These confer an enzymatic activity to the protein that converts α-KG producing a new oncometabolite D-2-hydroxyglutarate (2-HG), found increased in mutant *IDH* cells[71]. This metabolite is an inhibitor of several α-KG-dependent deoxygenase reactions, for it is a cofactor for proteins of the TET family and the JumonjiC domain histone demethylases (erasers of DNA and histone methylation). One of these enzymes is TET2. By inhibiting TET2 the molecular alterations in *IDH* give rise to the strong hypermethylation phenotypes found in *IDH* mutant AML patients[1,37], which results in undifferentiated states. On the other hand, by inhibiting the JumonjiC type enzymes it deregulates histone methylation. Accordingly, a general increase in certain histone methylation marks has revealed closed DNA conformation in *IDH1*-KI mouse cells[80] and *IDH* mutation induce CNS-derived[71].

1.4 Epigenetic patterns in AML

> *"DNA methylation and its influence on gene expression are key in understanding cancer pathogenesis. Even though it is becoming clear that DNA methylation strongly interacts with other components of the epigenetic machinery such as histone modifications, aberrant DNA methylation can still be regarded as a crucial hallmark of cancer by itself."*[81]

Biology has evolved a great deal in grasping the rules of gene expression by recognizing that the process involves several dimensions of regulation. The first reports described heritable DNA methylation, histone modifications and micro RNA variability. Nowadays, epigenetics refers to all heritable changes in gene expression that are not due to any alteration in the DNA sequence, this includes DNA methylation, chromatin remodeling (by histone modifications or binding of certain proteins to DNA, like the insulator binding CTCF) and the activities of non-coding RNAs. The interplay between these epigenetic components has not been completely determined, but a complex picture of gene expression control has emerged. Since the correct distribution of DNA methylation is essential for every tissue differentiation and homeostasis[82] deregulations of epigenetic processes are connected to oncogenesis. DNA methylation is nowadays recognized as a mechanism by which cells acquire cancer hallmark capabilities[83] (of undifferentiating and self-renewal), playing important roles in initiation, progression and maintenance of malignant phenotypes.

Since the patterns of aberrant gene expression in AML are not well explained by the classifications based on mutational patterns the role of DNA methylations has been under examination. The quantification of 5-cytosine methylation (5-methylcytosine, m^5C) in the cytosine-phosphate-guanine sequential nucleotides (CpGs) based in genome-wide techniques has provided valuable information about observed gene expression patterns in several cancers including colon, lung and breast cancer[84] and was expected to do the same for AML.

Genome-wide analysis of DNA methylation suggested that aberrant DNA methylation in cancer occurs at defined genomic locations, termed cancer-specific differentially methylated regions (DMRs)[84]. Genome-wide studies indicated that cancer cells have global DNA hypomethylation when compared to their respective tissues,

with regions of hypermethylation associated with CpG-islands at promoter sites[84,85], creating patterns that could even guide us into better clinical classifications of tumors[86].

Standing in contrast to these solid tumors, AML has a more complex picture of epigenetic regulation. The earliest analyses of DNA methylation in AML found aberrant DNA methylation in the promoters of some genes, for the available data covered only 14,000 promoters[87,88]. Later, a crucial analysis of multidimensional data in AML integrated genetic and epigenetic patterns of 344 AML samples and defined 16 subclasses of patients according to their DNA methylation profiles using 50,000 CpGs accessed by HELP (HpaII tiny fragment enrichment by ligation-mediated PCR)[89]. These 16 groups of patients presented unique methylation patterns when compared to normal bone marrow $CD34^+$ cells. Unexpectedly most groups were largely hypermethylated and only a minority hypomethylated, additionally many promoter regions were actually hypomethylated[89].

This diversity of epigenetic changes in AML adds complexity to the already complex genetic heterogeneity. The only common epigenetic signature of these AML samples consisted of 45 genes differentially methylated (mostly hypermethylated in relation to the normal bone marrow $CD34^+$ cells) and that was obtained considering only 70% of patients[89]. Other studies, that profiled the methylation status of more patients and more genes, also found only small sets of genes had methylation patterns deregulated across samples[37,90].

In contrast, most studies find groups of patients that are reflective of the presence of specific cytogenetic and genetic aberrations in samples. Namely, there were different groups for the fusions *PML-RARA*, *CBFB-MYH11, RUNX1-RUNX1T1* (*AML1-ETO*) and then for the mutations of *CEBPA* double mutants and *NPM1* and *CEBPA* silenced in one study[89]; and in another study for *MLL* translocations vs *IDH* mutated samples[90].

More recently the epigenetic landscapes of AML have been analysed by TCGA, using the genome-wide DNA methylation BeadChip from Illumina® covering 99% of RefSeq genes with over 450,000 CpGs. In this study TCGA determined significant changes in methylation at 160,516 CpGs loci when the 192 patients were compared to $CD34^+CD38^-$ cells from healthy donors, confirming hypermethylations were the majority of changes (67%) while hypomethylations were only present in a minority of places (33%)[37]. In this case, groups of samples with specific genetic lesions were still present but not so clearly defined[37], with the strongest methylation signatures being obtained using regions of the genome with low amounts of CpGs. This brought back

the questions about which CpGs might be more important for regulation of gene expression.

The latest study followed indications that the defining DMRs in leukemia might not be the typical hypermethylations in gene promoters and instead be correlated to regulatory elements outside of promoters[91]. These would possibly be in regulatory DNA sequences, which in vertebrates often have little or no methylation, like enhancers[92]. They used a new technique not centered on promoters and denominated enhanced reduced representation bisulfite sequencing (ERRBS), that allowed for the widest probing of the methylation status so far (about 951,000 CpGs). Using this dataset they found that despite the methylation heterogeneity of samples some genetic lesions could still be correlated to specific DNA methylation profiles (Figure 1.4.1)[91].

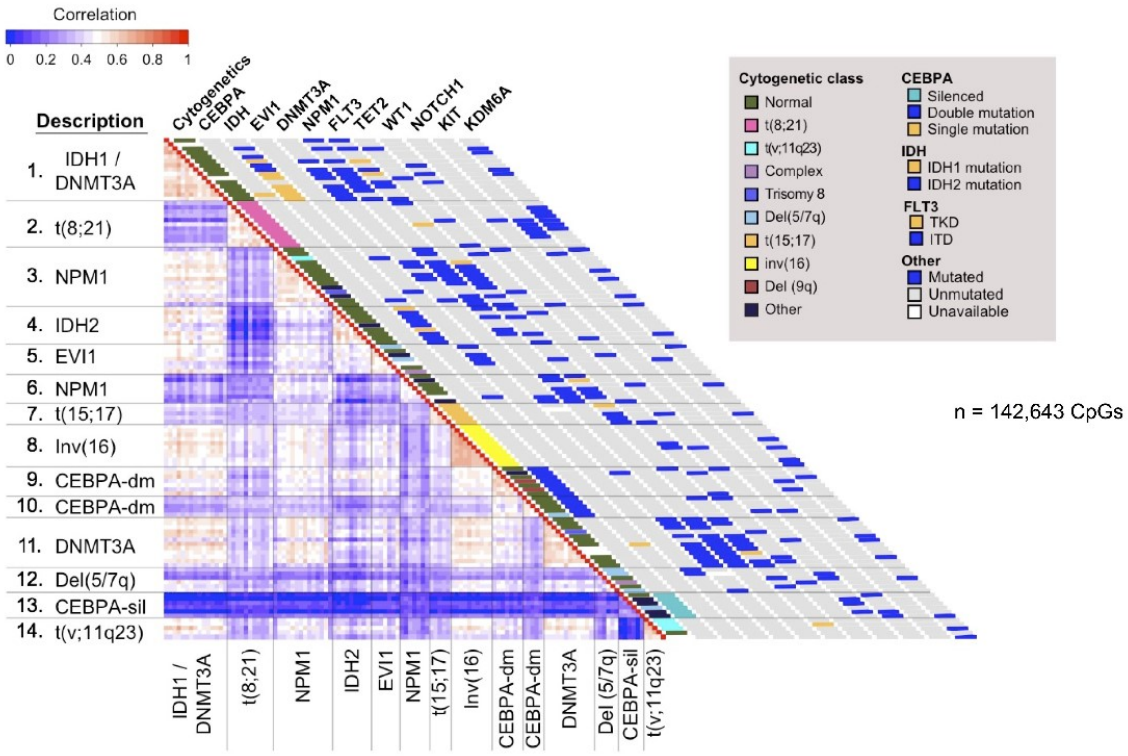


Figure 1.4.1 Epigenetically defined classification of AML.
"Representation of hierarchical clustering results based on ERRBS data using a correlation matrix heatmap. ERRBS defines AML subtypes with distinct molecular and cytogenetic characteristics. Groups were defined using a hierarchical clustering approach and labeled according to their dominant distinguishing molecular and cytogenetic features. The lower triangular heatmap represents the correlation between the most divergent CpGs represented in all samples. Cytogenetic and specific molecular features are represented in the diagonal bars on the right." Adapted from Glass *et al.*[91].

Additionally, the epigenetic identity of genetically similar samples was best captured by using gene CpGs in gene neighborhoods (between 2kb and 50kb away

from the transcription start site or transcription end site) rather than promoters, or by CpGs in shores in opposition to CpGs islands if using only CpGs in promoters[91].

These high-throughput DNA methylation data in AML were not only mere curiosities, due to its dynamic nature it provides potential targets for therapy. Knowing that AML is populated with DNA hypermethylation provided bases for the application of hypomethylating agents in clinical settings, although their mechanisms of action are not well defined since they indiscriminately remove methyl groups from DNA[62]. Therefore, targeting specific genes with expression affected by different DNA methylation marks would possibly be more effective.

Many studies have found that DNA methylation could predict clinical outcome in AML patients and therefore indicated aberrant DNA methylation can serve as a biomarker for risk stratification (Table 1.4.1).

Table 1.4.1 Prognostic genes regulated by DNA methylation.

Genes with differential DNA methylation that were identified by genome-wide detection methods applied to large cohorts of AML patients. Adapted from Li *et al*. 2017[93].

Reference	Method	AML group	Prognostic genes regulated by DNA methylation
Figueroa *et al.* [89]	HELP	344 *de novo* AML, median Age 48y (range:15-77)	*BLR1*[*CXCR5*]*, BTBD3, E2F1, FAM110A, FAM30A, GALNT5, KIAA1305, LCK, LMCD1, PRMT7, SLC7A6OS, SMG6, SRR, USP50, VWF, ZFP161*
Li *et al.* [94]	ERRBS	138 Paired AML (diagnosis and relapse), mean Age 50.5±14y	*CCDC85C, CHL1, ELAVL2, FAM115A, FAM196A, GPR146, GPR6, HEL Z2, ID4, IL2RA, KCNG3, LOC254559, LOC284801, NPAS2, PCDHAC2, PROB1, SHISA6, SLC18A3, SOCS2, TRIM67, ZFP42*
Marcucci *et al.* [95]	MethylCap -seq	134 CN-AML training (Age range ≥60y) + 355 CN-AML validated (Age range 17-83y)	*AATK, ACAP3, ADCK2, ADCY6, AGPAT9, AHCY, ALOX15B, ANXA6, APBB1, APOD, AQP11, ARHGAP27, AXL, BRF1, C15orf62, C17orf77, C8orf51, CABLES1, CARD11, CD34*, CHMP7, CISH, CLDN15, CLEC3B, DDIT4, DHCR24, DHRS12, EGFL7, ETS1, EVC, F2RL1*, FAM92A1*, FCHO1, FKBP4, FLVCR1, FLVCR1-AS1,FZD6, GAL3ST3, GCNT2, GIT1, GPR56, H1F0, HCN2, HIVEP3, IQSEC1, KCNK6, KDM2B, KLHL3, KNCN, LOC646627, MDFI, ME3, MEOX1, MIR126, MIR155HG*, MVD, NAV1, NBL1, NLRP1, PLK3, PMM1, PRKCZ, PRKG2, RAB36, RGS3, RHOC*, RHPN1, SCARF1, SCRN1*, SH3TC1, SPRY1, SRC, TBL2, TCEA3, TENC1, UBXN6, VWA8*, WDR16, WDR86, WRAP53, ZNF623, ZNF70*
*Seven genes (*CD34, RHOC, SCRN1, F2RL1, FAM92A1, MIR155HG,* and *VWA8*) had not only DNA methylation regions (DMRs) but also expression levels that were associated with outcome.			

Unfortunately, even when analysing many CpGs, studies usually did not find many genes with DNA methylation signatures associated with prognosis. Only 16, 22 and 82 different genes were found to be correlated to survival, by the studies of Figueroa *et al.*[89], Li *et al.*[94] and Marcucci *et al.*[95] respectively. There was no overlap between them, which is understandable because the cohorts used have very different compositions in terms of AML genetic and clinical factors. The impossibility of comparison is made worse by the fact that these investigations were done using

different techniques. In addition, these predictive signatures were still not reproduced by independent studies.

Therefore, to establish DNA methylation levels that could be used as prognostic indicators has so far been challenging. However, the possibility of clarifying the driving forces of gene expression alterations from the DNA methylation changes in AML is still very attractive and is the subject of continuous study. Establishing the prognostic value of individual DNA methylation sites as biomarkers, especially on the context of specific cytogenetic subgroups, is a very appealing prospect since it could lead to new therapeutic strategies.

1.5 Elderly AML specificities

> *"First, how does the hematopoietic system change with age, how does this lead to a spectrum of hematological conditions, and would it be possible to pharmacologically intervene in this process? Second, if hematological disease presents in an elderly patient, should this be treated differently than if it had occurred in a young patient?"*[3]

These are pertinent questions for which answers have so far eluted hematologists. They are of utmost importance because AML is primarily a disease of the elderly population with the incidence rising with age. According to the SEER population statistics, only 1.9 of 100,000 people under 65 years old develop AML, a number rising to 19.1 for the elderly population of 65 years and older (between 2009 and 2013)[96]. Expectedly, the median age of patients with AML at the time of diagnosis is around 71 years old (indicated by the compulsory reports of population-based studies like the Swedish AML registry with about 3,363 adult AMLs)[46,45,97].

1.5.1 Survival of elderly AML patients

Overall, elderly patients with AML have a poor survival probability, as studies show that survival to AML decreases with advanced age (Figure 1.5.1)[46,45,98].

Although the goal of curing AML can only be reached using intensive chemotherapies, the therapy choices for older patients also include less intensive chemotherapy or supportive care. This depends on the doctors' perception of the patient's condition (performance status, ECOG score, co-morbidities, and tolerance to intensive chemotherapy). These patients are many times considered *unfit* for the intensive chemotherapy for fear of the high rates of early death in initial treatment. For instance, patients older than 75 years with an ECOG performance score of 3 (*"capable of only limited self-care; confined to bed or chair more than 50% of waking hours"*) or higher have a 30-day mortality rate of more than 50%[44].

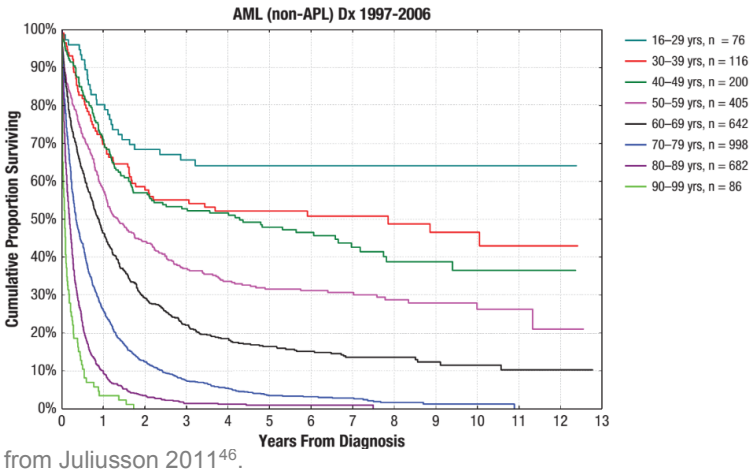


Figure 1.5.1 Survival of patients with AML in a population study according to their age at the time of diagnosis. Kaplan-Meier curves for all the patients diagnosed with AML (non-acute promyelocytic leukemia) between 1997 and 2006, treated and untreated (n=3205). Divisions were done by decades. With data used was from the compulsory Swedish registry of AML with a median follow-up of 6.2 years. Adapted from Juliusson 2011[46].

As a result, in the US 60% of AML patients 65 years or older have not received therapy 3 months after diagnosis[99]. The lack of viable options leads clinicians in Europe to widely use the less intensive approaches in patients over 65 years old, like decitabine or azacitidine, for although the effectiveness of these hypomethylating agents is not well established, they are promising[100,101]. They remain a viable option, having in mind that even with intensive chemotherapy only 50% of patients older than 60 years achieve complete remissions and 50% of those would have relapsed within 2 years[102].

Advances in therapy in the elderly patients have been compromised by extrapolation of results from clinical trials that mostly use young patients (frequently younger than 60 years). Elderly patients tend to be excluded from clinical trials for many reasons. The selection bias is frequently due to strict criteria for entering trials with intent-to-treat and by the drop out due to the impossibility of many of this patients to begin treatments, to complete them, or to go through consolidation phases[103].

Therefore, we know little about how and why treatments *work* or *not work* in the elderly group of AML patients. Higher rates of resistance to intensive chemotherapy treatment regimens and high rates of relapse have been observed in the older patients when compared to younger AML patients. Response rates were observed to decrease with age (as well as the median overall survival) even when there were no protocol-directed differences in dosing of patients[44]. Moreover, in a recent study, the treatment response of patients 57-59 years old to a full-dose chemotherapy proved to be no different from 60-63 year's patients receiving an attenuated-dose therapy[104]. Therefore it is regularly concluded that age is an important independent determinant of therapy-response in AML.

1.5.2 Molecular characteristics of elderly AML

These facts have been attributed to several disease characteristics displayed by elderly AML when compared to young AML patients, like the fact that older patients are more likely to have adverse cytogenetics[105,106]. Nevertheless, elderly AML patients irrespective of their cytogenetic group show an inferior outcome compared to younger patients with the same risk alterations[106,107].

In an effort to examine the phenotype of this disease a study analysed the status of oncogenic pathways in elderly AML in comparison to younger AML[108]. They found older patients had a higher probability of RAS, Src, and tumor necrosis factor (TNF) pathway activation[108].

All of these data underscored the biological differences between AML in younger and older patients. Evidences that led us to postulate that elderly AML is a different entity for which a targeted treatment is needed and should be properly investigated. As it remained an open question if additional molecular alterations have important contributes to the poor prognosis of AML in the elderly patients this was one focus of our project[1].

Meanwhile, attending to these indications in last two years some studies that sequenced genes known to be frequently mutated in AML have made a point in dividing their cohorts into young and elderly patients to look at frequencies of genetic alterations[34, 38,40].

In general, it is now established the commonly mutated genes in older patients with AML included *RUNX1*, *TET2*, *IDH2*, *ASXL1*, *SRSF2*, *TP53*, *BCOR* and *SF3B1* while *WT1* mutations were more frequent in the younger patients[1,34,38,40].

Since the genetic profiles might explain some of the poor prognosis of elderly AML the origin of this very specific profiles should provide important information for new treatment design.

As age-related changes of the diseased hematopoietic system are found they must be the result of the aged state of the HSCs. Therefore we should explore the possible effects of age on the progenitor cells that give rise to the elderly AML.

CHAPTER 2. AIM OF THE PROJECT

Many studies of AML pathogenesis in elderly people led us to raise the hypothesis that, **AML in the elderly is not just AML in an older person**. Is the landscape of molecular alterations in AML different in older patients with a higher load of pre-existing alterations? Which pathogenic processes are more affected in elderly AML than in younger patients? Do specific genetic and epigenetic features contribute to the poor prognosis and treatment resistance of elderly cohorts?

Though complex patterns of molecular alterations had been characterized in some younger cohorts, comprehensive and integrated studies (genetic and epigenetic) in older cohorts were lacking. We aimed to find the **genetic alterations and global epigenetic changes** in elderly AML that differed from AML in younger patients. We expected both profiles were necessary to define the specificity of elderly AML.

1. We first concentrated on obtaining a picture of the **relevant molecular alterations** in this tumor entity:
   - ✓ Exploring recurrent genetic alterations known in AML in a large number of patients with elderly AML using targeted NGS.
   - ✓ Probing for unknown events, analysing a broad set of candidate genes by an extended NGS panel of 555 genes.
   - ✓ Describing global genome methylation patterns for elderly AML using an array that covers over 450,000 methylation sites.
2. We aimed to define **elderly AML** comparing its genetic and epigenetic patterns to profiles of younger AML cohorts.
3. We next wanted to determine the **pathways/cellular processes** that are more affected in elderly AML than in younger AML cells (both by mutations and/or epigenetic alterations).
4. We postulated the **interconnection of genetic and epigenetic profiles** would allow the division of elderly AML patients in epigenetically defined subgroups associated with specific genetic alterations.
5. We meant to explore possible genetic/epigenetic **factors responsible for the poor survival** of elderly AML and therefore reasons for the *age* factor.

We expected the knowledge of this distinct genomic/epigenomic entity and its complexity to lay a foundation for future development of **smarter therapeutic regimes**.

CHAPTER 3. PATIENTS AND METHODS

3.1 Elderly AML cohort characteristics

The cohort studied was composed of 93 AML patients enrolled in the Study Alliance Leukemia (SAL) registry between 2005 and 2014. All patients provided written informed consent in compliance with the Declaration of Helsinki. This cohort consisted of patients 65 years and older at the time of AML diagnosis (Figure 3.1.1), chosen using only this criteria and the availability of sufficient material in quality and quantity as well as clinical information[1]. Clinical data are summarized in Table 3.1.1, showing our cohort had a composition within expected, due to similarities to reports of AML in general population[97], where 75% were diagnosed with *de novo* AML, 19% sAML and 5% therapy-related AML (further detailed in Appendix A).

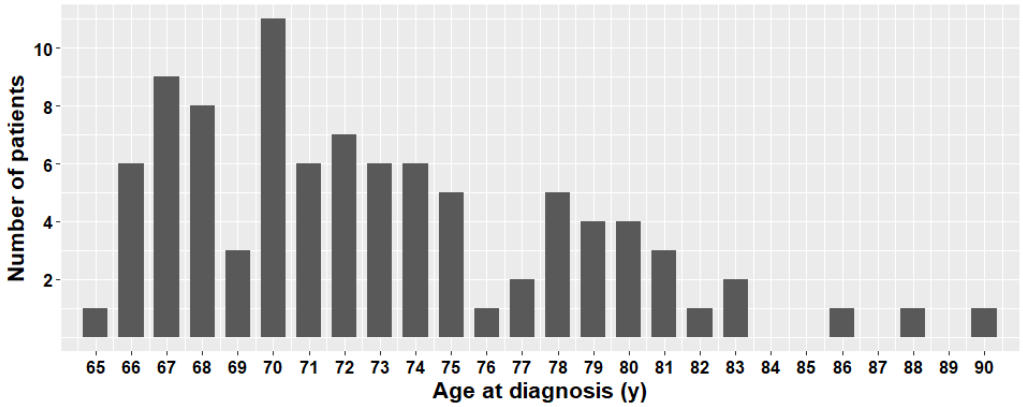


Figure 3.1.1 Distribution of patients' age at diagnosis in the SAL elderly AML.
The plot shows the number of samples present in the SAL cohort for each specific patient age, in intervals of 1 year, calculated from the date of birth.

Table 3.1.1 Characteristics of elderly AML cohort.

Summary of characteristics of patients: *de novo* AML (no previous leukemic related diagnosis), secondary AML (sAML, with a previous diagnosis of myelodysplastic syndrome or myeloproliferative neoplasm), or therapy-related AML (t-AML, patients with prior exposure to cytotoxic therapy and/or radiotherapy). The cytogenetic groups were defined as: cytogenetically normal (CN, no detected cytogenetic abnormalities), core binding factor (CBF, includes t(8;21) or inv(16)/t(16;16)), Complex (3 or more cytogenetic abnormalities), or other (1 or 2 cytogenetic abnormalities). Treatments of patients were divided according to the application of standard intensive chemotherapy or not (the latter consisted of palliative or best supportive care, which includes treatment with hypomethylating agents). Several abbreviations were used: *FLT3* internal tandem duplications (*FLT3*-ITD), mutations in nucleophosmin altering tryptophan residues in the C-terminus of the protein (*NPM1c*), white blood cell counts (WBC), Lactate dehydrogenase (LDH), bone marrow (BM), months (mon) and years (y). As presented in Silva *et al.* 2017[1].

Clinical characteristics, n=93	
Age at diagnosis (y), median (ranges)	72 (65-90)
Sex (Male), no. (%)	47 (50.5)
***de novo* AML, no. (%)**	70 (75)
sAML, no. (%)	18 (19)
t-AML, no. (%)	5 (5)
LDH (U/L), median (ranges)	406 (138-4219)
WBC (/nL), median (ranges)	14.8 (0.9-210)
BM Blasts, % (ranges)	75 (25-99)
Cytogenetic, n=75	
CN, no. (%)	28 (37)
CBF, no. (%)	5 (6.7)
Complex, no. (%)	12 (16)
Other, no. (%)	30 (40)
Molecular data	
***FLT3*-ITD, no./ total no.(%)**	15/70 (21)
***NPM1c*, no./ total no.(%)**	15/69 (22)
Treatment, n=90	
Palliative or best supportive care, no. (%)	19 (21)
Intensive chemotherapy, no. (%) [no. stem cell transplantation]	71 (79) [10]
Survival, n=92	
Median survival time (mon)	15
Follow up time (mon), median (ranges)	7.4 (0.1-92)

3.2 Mutations in protein coding sequences of 555 genes

We obtained sequence data from target enrichment coupled NGS of 555 genes (Table 3.2.1).

DNA was extracted, using the AllPrep DNA/RNA kit (©QIAGEN, N.V.), after a Ficoll isolation (Ficoll-Paque™ PLUS, GE Healthcare, USA) for enrichment of the fraction of blast cells from the bone marrow aspirates of 93 patients (or from peripheral blood mononuclear cells, n=1) obtained prior to treatment.

For the exome capture, SureSelect^{XT} custom 0.5Mb-2.9Mb was used to prepare a library from sonicated DNA with lengths of 300-400bp (SureSelect®, Agilent Technologies, USA) according to manufacturer's protocol, using 1µg to 3µg of DNA as starting material according to availability. During this preparation, DNA samples were analysed using an Agilent DNA 1000 chip run on an Agilent Bioanalyzer 2100 (G2938B, Agilent Technologies, USA) to assess sizes of DNA fragments obtained after sonication procedure (average lengths of 280bp). Similar electropherogram summaries were made for the final libraries of captured fragments of all samples in order to assure fragment sizes averaging between 300-330bp. For confirmation of specific capture of the targets in the DNA libraries (obtained for each patient), two simultaneous qPCR reactions were performed. A positive control verifying the presence of exon 23 of *DNMT3A* (labeled probe FAM and BHQ1) and a negative control verifying the non-hybridization of the gDNA of the non-target *GATA4* using a set of primers that span the promotor (labeled probe Joe and TAMRA). The DNA amount of the samples was then quantified using Quantus™ Fluorometer (Promega Corporation, USA) to guarantee the normalized amount of 16 patients per pool (10nmol/L of each patient sample).

The captured targets were subjected to massively parallel sequencing using Illumina HiSeq2000 with 2x100bp paired-end reads (functional genome analysis gene center, Muenchen), using 1 pool per sequencing lane. Sequenced reads were aligned using a BWA to the GRCh37/hg19 reference genome. From the 25±10 million reads per sample retrieved about 91.8±8.2% of reads mapped to the genome, about 99.5±0.4% of mapped reads had high-quality scores (Q≥20, Figure 3.2.1). After removal of PCR duplicates, we obtained a median target coverage of 408±164 reads and then considered only targets sequenced with the minimal 30x coverage (98.4±1.8% of all targets, Figure 3.2.1). Non-synonymous single nucleotide variants, small insertions or deletions in the coding sequence and affecting splice sites were detected with VarScan2 and Pindel according to standard quality assurances.

Mutations were considered if alterations were present at a variant allele frequency (VAF) higher than 20%, which was further adjusted for the genes in the X chromosome and therefore denominated adjusted VAF.

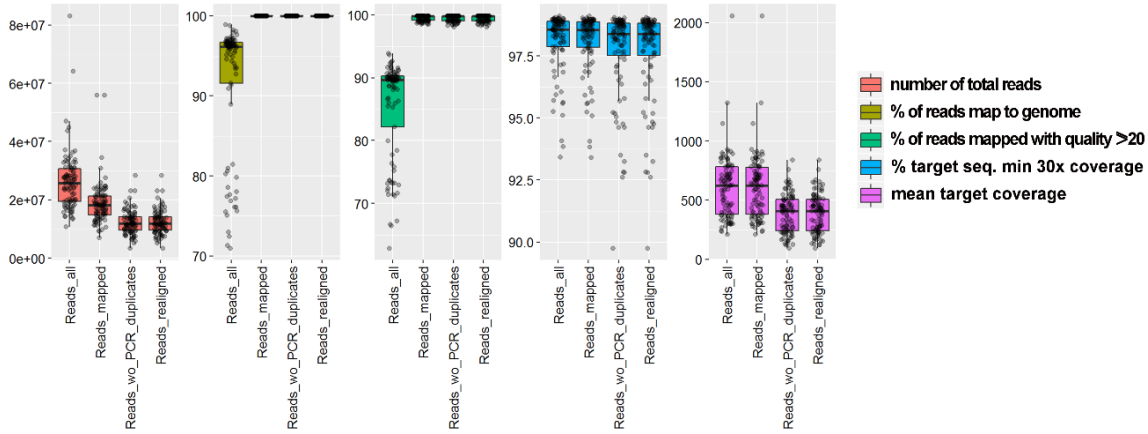


Figure 3.2.1 Quality controls of mapping and coverage of the target sequencing of the SAL elderly AML samples.

The plots show results of the application of the BWA algorithm, used for alignment of NGS reads to the genome. In the horizontal axis are the points at which data was collected during the mapping process (before and after alignment, after removal of PCR duplicates and the final trimmed/realigned fragments). In the vertical axis are the numbers of reads or the % of reads obtained for each of the 93 samples, according to the analysis reported in the legend. Abbreviations in legend and axis were seq. for sequences, min for minimum, wo for without and 30x for 30 times.

Table 3.2.1 Mutated genes selected for the targeted NGS design.

List of the 555 genes previously correlated to leukemia or other cancer types and developmental diseases. Listed in alphabetic order.

ABI1	BUB1B	CARD11	EZH2	HIP1	LCP1	NBN	PIM1	SEPT5	TIRAP
ABL1	CCDC6	CARS	EZR	HIPK1	LHFP	NCKIPSD	PLAG1	SEPT6	TLX1
ABL2	CCNB1IP1	CASC5	FAM46C	HIST1H4I	LIFR	NCOA1	PML	SEPT9	TLX3
ACACA	CCND1	CBFA2T3	FANCA	HLF	LMNA	NCOA2	PMS1	SET	TMPRSS2
ACER1	CCND2	CBFB	FANCC	HMGA1	LMO1	NCOA4	PMS2	SETD2	TNFAIP3
ACSL3	CCND3	CBL	FANCD2	HMGA2	LMO2	NDRG1	POM121	SF3B1	TNFRSF14
ACSL6	CCNE1	CBLB	FANCD2OS	HNF1A	LPP	NEBL	POU2AF1	SFPQ	TNFRSF17
ACTN4	CCNL1	CBLC	FANCE	HNRNPA2B1	LRIG3	NF1	POU5F1	SH3GL1	TNRC18
AFF1	CD274	DAB2IP	FANCF	HOOK3	LYL1	NF2	PPARG	SH3GL1	TOP1
AFF3	CD74	DACH1	FANCG	HOXA11	MAF	NFE2L2	PPARGC1A	SIRT1	TP53
AFF4	CD79A	DAXX	FAS	HOXA13	MAFB	NFIB	PPP2R1A	SIRT3	TPM3
AKAP9	CD79B	DCP1A	FAT1	HOXA9	MALT1	NFKB2	PRCC	SLC34A2	TPM4
AKT1	CDC42EP1	DCPS	FAT2	HOXC11	MAML2	NIN	PRDM1	SLC45A3	TPR
AKT2	CDC73	DDB2	FAT3	HOXC13	MAP2K4	NKX2-1	PRDM16	SLCO1B3	TRDMT1
ALDH2	CDH1	DDIT3	FBXO11	HOXD11	MAPRE1	NONO	PRF1	SMAD4	TRIM24
ALK	CDH11	DDX10	FBXW7	HOXD13	MDM2	NOTCH1	PRG4	SMAP1	TRIM27
AMER1	CDK12	DDX5	FCGR2B	HRAS	MDM4	NOTCH2	PRKAR1A	SMARCA4	TRIM33
APC	CDK4	DDX6	FCRL4	HSP90AA1	MDS2	NPM1	PRKCZ	SMARCB1	TRIP11
APOB	CDK6	DEK	FEV	HSP90AB1	MED12	NR4A3	PRRX1	SMO	TSC1
ARHGAP26	CDKN2A	DICER1	FGFR1	HSPB2	MEN1	NRAS	PSIP1	SNX29	TSC2
ARHGEF12	CDKN2C	DLL1	FGFR1OP	IDH1	MET	NRIP3	PTCH1	SOCS1	TSHR
ARHGEF17	CDX2	DNM2	FGFR2	IDH2	MITF	NSD1	PTEN	SORBS2	TTL
ARID1A	CEBPA	DNMT1	FGFR3	IGF1R	MKL1	NTRK1	PTPN11	SOX2	U2AF1
ARID2	CEBPD	DNMT3A	FH	IGFBP7	MLF1	NTRK3	PTPRT	SPECC1	UBE4A
ARNT	CELA3B	DNMT3B	FHIT	IKZF1	MLH1	NUMA1	RABEP1	SRGAP3	UCP2
ASPSCR1	CHCHD7	DUX4	FIP1L1	IL2	MLL	NUP214	RAD21	SRSF2	UCP3
ASXL1	CHEK2	E2F1	FLCN	IL21R	MLL2	NUP98	RAD51B	SRSF3	USP6
ATF1	CHIC2	EBF1	FLI1	IL6ST	MLL3	NUTM1	RAF1	SS18	VAV1
ATIC	CHN1	ECT2L	FLNA	IL7R	MLLT1	NUTM2A	RALGDS	SS18L1	VHL
ATM	CIC	EED	FLT3	ING1	MLLT10	OLIG2	RANBP17	SSX1	VTI1A
ATRX	CIITA	EEFSEC	FNBP1	IRF4	MLLT11	OMD	RANBP2	SSX2	WAS
AUTS2	CLP1	EGFR	FOXL2	ITK	MLLT3	P2RY8	RAP1GDS1	SSX4	WHSC1
BAALC	CLTC	EIF4A2	FOXO1	JAK1	MLLT4	PAFAH1B2	RARA	STIL	WHSC1L1
BAP1	CLTCL1	ELF4	FOXO3	JAK2	MLLT6	PALB2	RB1	STK11	WIF1
BCL10	CNBP	ELK4	FOXO4	JAK3	MN1	PATZ1	RBM15	STRN3	WLS
BCL11A	CNOT	ELL	FOXP1	JARID2	MNX1	PAX3	RCSD1	SUFU	WRN
BCL11B	CNR2	ELN	FRYL	JAZF1	MPL	PAX5	RECQL4	SUZ12	WT1
BCL2	COL1A1	EML4	FSTL3	JUN	MSH2	PAX7	REL	SYK	WWTR1
BCL3	COX6C	EP300	FUBP1	KDM5A	MSH6	PAX8	RET	TAF15	XPA
BCL6	CREB1	EPOR	FUS	KDM5C	MSI2	PBRM1	RHOH	TAL1	XPC
BCL7A	CREB3L1	EPS15	GAS7	KDM6A	MSN	PBX1	RMI2	TAL2	XPO1
BCL9	CREB3L2	ERBB2	GATA1	KDR	MTCP1	PCM1	RNF213	TCEA1	YWHAE
BCOR	CREBBP	ERC1	GATA2	KDSR	MTOR	PCSK7	ROS1	TCF12	ZBTB16
BCORL1	CRLF2	ERCC2	GATA3	KIAA0284	MUC1	PDCD1LG2	RPL22	TCF3	ZFYVE19
BCR	CRTC1	ERCC3	GMPS	KIAA1549	MUTYH	PDE4DIP	RPN1	TCF7	ZMPSTE24
BECN1	CRTC3	ERCC4	GNA11	KIF3B	MYB	PDGFB	RUNX1	TCF7L2	ZMYM2
BIRC3	CRYAB	ERCC5	GNAQ	KIF5B	MYC	PDGFRA	RUNX1T1	TCL1A	ZNF276
BLM	CTNNB1	ERG	GNAS	KIT	MYCL	PDGFRB	SBDS	TCL6	ZNF331
BMPR1A	CXCR7	ETV1	GOLGA5	KL	MYCN	PER1	SCD	TET1	ZNF384
BRAF	CYLD	ETV4	GOPC	KLF6	MYD88	PHF13	SDC4	TET2	ZNF521
BRCA1	C15orf65	ETV5	GPC3	KLK2	MYH11	PHF6	SDHAF2	TFE3	ZRSR2
BRCA2	C20orf112	ETV6	GPHN	KRAS	MYH9	PHOX2B	SDHB	TFEB	
BRD3	C2CD3	EVI1	H3F3B	KTN1	MYO1F	PICALM	SDHC	TFG	
BRD4	C2orf44	EWSR1	HERC1	LAMC3	MYST3	PIK3C2B	SDHD	TFPT	
BRIP1	CAMTA1	EXT1	HERPUD1	LASP1	MYST4	PIK3CA	SEPT11	TFRC	
BTG1	CANT1	EXT2	HEY1	LCK	NACA	PIK3R1	SEPT2	THRAP3	

3.3 Sequence data analyses, variant classification and confirmations

Variants were functionally annotated using Ensemble[109]. To differentiate known and putative leukemia driver mutations from known germline polymorphisms, all variants listed in the National Center for Biotechnology Information Short Genetic Variations database (dbSNP, version 138)[110] were eliminated. We also eliminated variants with SNP annotation from version 142 that were not reported in the catalogue of somatic mutations in cancer (COSMIC)[111]. We then included variations in positions confirmed to have somatic mutations from all TCGA cohorts[112] with a non-tumor control sample (Acute Myeloid Leukemia, Ovarian serous cystadenocarcinoma, Rectum adenocarcinoma, Colon adenocarcinoma) and variants that were labeled pathogenic. Finally, all variants presented have a minor allele frequency (MAF) <0.01 according to the 1,000 genomes project[113].

Artifacts of the sequencing procedure were analysed by submitting to the same NGS procedures 9 DNA samples from the bone marrow of healthy donors and 5 DNA samples collected from AML patients in remission. For bone marrow samples of healthy donors, mononuclear cells were isolated using Ficoll (Ficoll-Paque™ PLUS, GE Healthcare, USA) and $CD34^+$ cells were then sequestered using CD34 Microbead Kit (MACS® Miltenyi Biotec, Germany), according to the manufacturer's instructions. Using the same protocol for DNA extraction, library preparation and analytical pipelines the data from these samples were retrieved. In this way, several molecular alterations deemed to be assay specific artifacts due to their recurrent presence in this samples (and high frequencies in the cohort samples >50%) were recognize and eliminated.

All remaining positions were manually curated in the Integrative genomics viewer[114,115] to guaranty provenience from trustable mapping. Genes that bear many fragments with noise or unknown mutations that were exclusively found in the tips of fragments were either investigated or straightly removed, depending on the quality of the mapping in the region.

Furthermore, we did several assessments of false discovery rate (FDR) by parallel techniques: using PCR followed by Sanger sequencing, routine diagnosis data and whole-exome data. We designed primer pairs for PCR amplification and sequenced 85 reactions with a Sanger technique (LGC Genomics GmbH) to check several uncertain mutations (either known or unknown). For example, we confirmed some mutations that are rare (*ASXL1* p.Gly646TrpfsTer12 and p.Ser689Ter, *APOB* p.Ser2429Leu, *DAB2IP* p.Thr11Ile; *EP300* p.Asn2209_Gln2213del, *ERCC2* p.Val476Ile and p.Gln662Ter; *FAT2* p.Asp1294Asn; *FAT3* p.Ile3265Thr; *MYST3*

p.Arg256Gln; *NSD1* p.Tyr1971Cys; *TET2* p.Arg544Ter, p.Arg1359Ser and p.Tyr1649Cys) and others unknown (*APC* p.Val569Met and p.Gln1303Lys, *APOB* p.Pro2620Ser, *BRCA2* p.Asp635Glu and p.Gln1595Pro, *FAT1* p.Ala4190Thr and p.Val4532Ile; *MYST3* p.Pro1003Ala; *NSD1* p.His390Gln; *TET2* p.Gln1652Ter, p.Gln1526Ter, p.Gly1391Val and p.Asn1387Ser). We also used Sanger sequencing to access the frequency of *NPM1c*, a known mutation in a place suspected to have suffered from bad coverage. In this case, 19 *NPM1c* were detected by sequencing 92 samples (Appendix A).

These refined filtering criteria had previously allowed us to keep true positive calls in a confidence of at least 95%[116]. After these methodologies, we were confident of having removed germline and unreliable mutations to keep FDR between 4-7%.

Finally, results were analysed with R software and Gitools[117]. Mutation data was deposited in NCBI's Sequence Read Archive (SRP089816).

3.4 Functional categorization of mutated genes

A protein functional interaction network was constructed with the Cytoscape 3.2.1[118], using the Reactome FIViz app[119] for gene set/mutation analysis to obtain a subnetwork of proteins affected (using genes mutated more than once and without linkers). The sub-network created was further clustered with a spectral partition based network clustering algorithm[120] into sub-networks (modules). Pathway and GO term enrichment analysis of the modules included modules with two or more genes, using FDR<0.05, on the several sources of pathway annotation available (KEEG, CellMap, Reactome, NCI PID, Panther, and BioCarta).

3.5 Correlation studies of mutated genes

Correlations between molecular alterations and/or other molecularly/clinically defined groups were calculated using Pearson correlation and significance was determined with Fisher exact tests or Mann-Whitney U test (for dichotomous and continuous variables respectively). The mutual exclusivity for gene pairs was analysed with CoMEt exact test[121] in R. Larger sets of genes patterns were analysed with Gitools[117] using the MutEx algorithm with 10,000 permutations.

3.6 Statistical analysis of overall survival

Overall survival (OS) analysis was performed in R using the survival package, with event defined as death by any cause or time censored at last follow-up. Differences between groups were tested with unadjusted Kaplan-Meier curves using

log-rank tests. Hazard ratios were estimated from Cox proportional hazard regression, used to evaluate the independent effects of covariates. Univariate, covariate and multivariate analysis were evaluated for Cox proportionality.

3.7 DNA methylation profiling

For DNA methylation assessment we used the Infinium® HumanMethylation450 BeadChip platform (Illumina Inc, CA, USA), reported to have high coverage (with 99% of RefSeq genes covered across gene regions and 96% of CpG islands that include shelves and shores). The array was performed on the 80 DNA samples from the SAL cohort with sufficient material available. A bisulfite–converted DNA was amplified, fragmented and hybridized to the probes of the microarray according to the manufacturer's protocol. For analysis of the Infinium® HumanMethylation450 BeadChip data we used the IDAT files with result values of 273 samples together (194 of the TCGA and 79 of SAL), or the 80 SAL samples only in R software, which were homogenized using the dasen function (wateRmelon package[122,123]). The DNA methylation fraction at each CpG is represented by the index of DNA methylation fraction, beta-values (β), calculated as $\beta = M/(M + U + \alpha)$, where M and U are methylated and unmethylated signal intensities and α is an arbitrary offset to account for measurements of low values. The dbSNP-related CpGs with MAF>0.01 were filtered out and the beta values of methylation in sex-related positions CpGs were removed.

Density profiles of the beta and M-values (β-logit2 transformed) samples were reviewed (Figure 3.7.1A-D). Principal components of all methylation values were analysed to guarantee the removal of batch effects and outliers (PCA, Figure 3.7.1E).

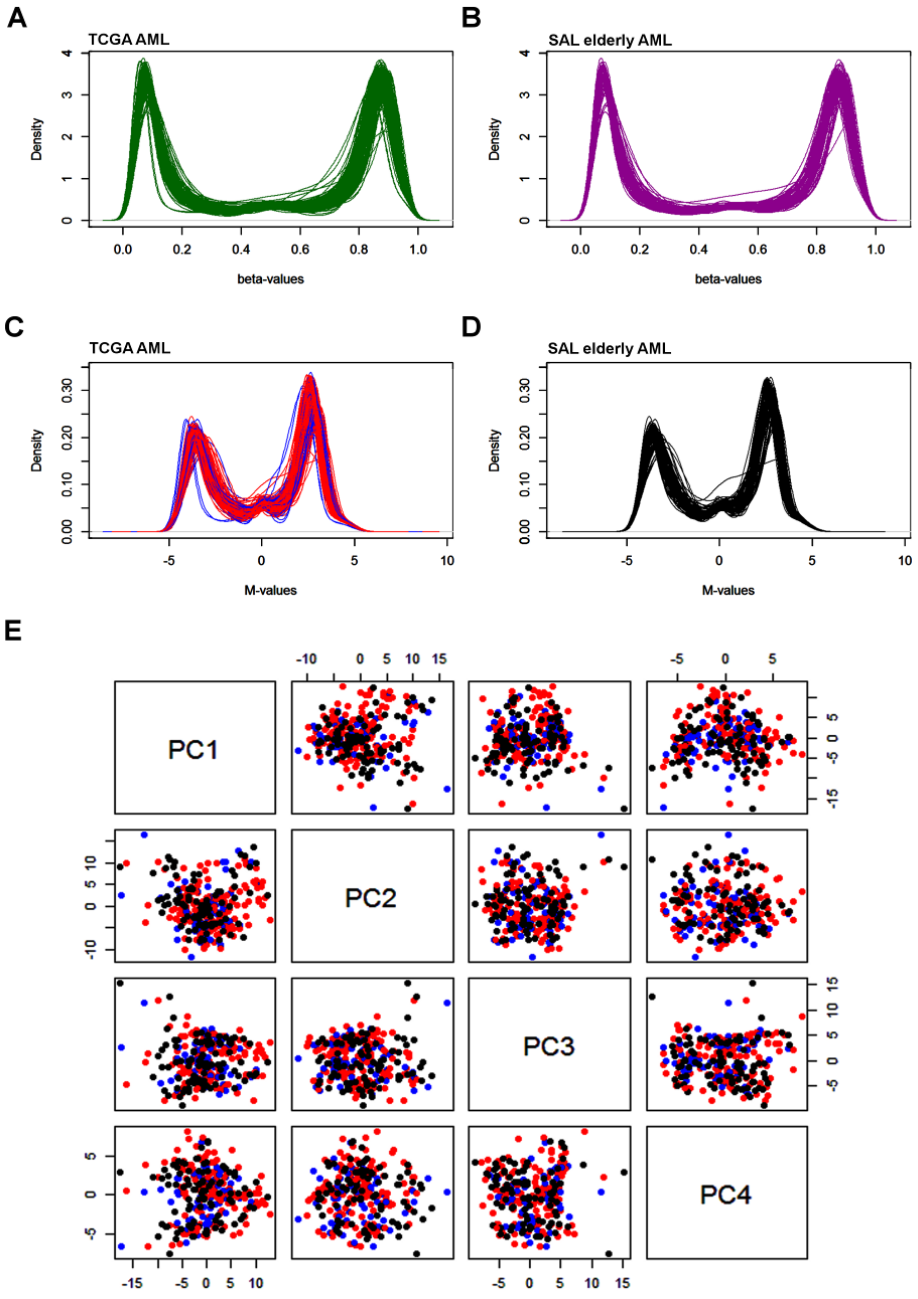


Figure 3.7.1 Representation of quality controls used for the integration of SAL and TCGA epigenetic data.

(A-D) Density plots that compare the distribution of methylation data from the TCGA and from the SAL cohorts. The first 2 graphics show β-values of the TCGA cohort (A, in green) and SAL cohort (B, in purple). The next 2 are representations of the corresponding M-values from the patients of TCGA cohort (C): young patients (less than 65y, in red) and elderly patients (65y or older, in blue) and from the SAL elderly cohort (D, 65y or older, in black). (E) Principal component plots for the first 4 components (which represent a good proportion of the variation). The plots show the homogeneity of the methylation data after application of the dasen function. Batch effects could not be perceived either for TCGA young cohort (less than 65y, in red), for TCGA elderly patients (65y or older, in blue) or for the SAL elderly cohort (in black).

Several unsupervised hierarchical clustering of the samples with M-values were done, using an average linkage of the Euclidian distance performed in Gitools[117]. Annotations of CpGs localized at promoters were done according to the Illumina manifest, as was the division of high-dense and low-dense CpGs regions (PHANTOM annotation) and the relative position of CpGs to CG islands (shelves and shores). A *t*-test was used to analyse the distribution of ages in the methylation groups defined by hierarchical clustering.

To determine differentially methylated regions (DMRs), we used R to apply 1000 permutations with the Bumphunter[124] algorithm and considered significant regions of pArea<0.05, family-wise error rate<10% and >2 CpGs differently methylated. The functional significance was extracted from the resulting cis-regulatory regions and genes which are proximal (within 5kb upstream and 1kb downstream) and distal (up to 1000kb) extracted from the entire genome GRCh37/hg19 by the Genomic Regions Enrichment of Annotations Tool v 3.0.0 (GREAT)[125]. These gene lists were analysed also using DAVID 6.7[126,127] and results were considered significant for EASE scores with FDR<10% and enrichment score of the respective cluster ≥1.3. Specific genes/regions were plotted in R using the heatmapByChromosome function from the methyAnalysis package. Calculation of the overlap of DMRs with genes, gene promoters and transcription start sites (TSS) were reported as analysed in EpiExplorer[128]. Positions of DMRs across the genome were plotted using the function plotKaryotype from the R package karyoploteR[129].

Array data was deposited and is accessible through NCBI's Gene Expression Omnibus (GSE86409).

3.8 Survival predictions from TCGA cohort gene expression profiles

We used the gene expression data from the TCGA cohort corresponding to the AML patients used in methylation. The TCGA RNA-seq dataset was used as processed by Pancan12[130], n=173, which is a log2-RSEM transformed and mean ranked matrix of 17,262 genes. The data were imported in R and for each gene a group of patients with gene expression lower than mean and another with equal or higher than mean was defined. The overall survival analysis performed was done following the implemented in the R package ‘survival’, with survival times given by time from diagnosis to the event (death from any cause) or censored at last follow-up. Differences between these groups were tested with unadjusted Kaplan-Meier curves using log-rank tests.

CHAPTER 4. RESULTS

4.1 Mutation profiles of the SAL elderly AML

We evaluated the mutations of a cohort of SAL elderly AML patients (n=93) with a median age of 72 years at diagnosis (65 to 90 years), using our self-designed panel NGS method for 555 genes. In total, we detected 814 mutations and considered putative somatic mutations in 281 of the 555 candidate genes (Figure 4.1.1, complete list of genetic alterations is in Appendix B).

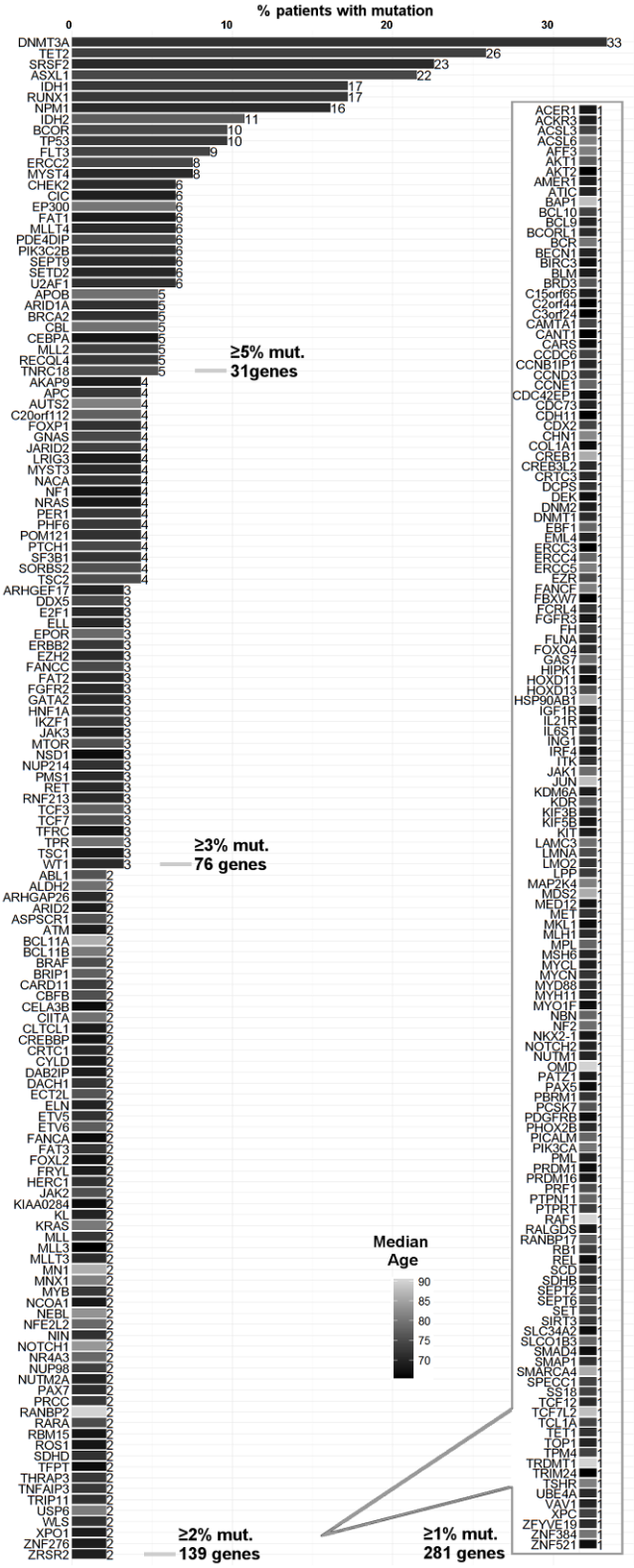


Figure 4.1.1 Frequencies of mutations in the SAL elderly AML for each gene.

The bar graph represents the percentage of patients with a mutation. The median age of the patients at diagnosis is depicted by a gradient code (in legend). A cumulative count of the number of genes with certain mutation frequencies is given in the right of the bars. Adapted from Silva *et al.* 2017[1].

A median number of 7 genes were mutated per patient (ranging 1 to 23 mutations per sample, Figure 4.1.2), most samples with at least one insertion or deletion and more than one SNV. We could not make comparisons of the rates of mutation in each patient to the reported average of 13 genes mutated per patient in the TCGA cohort. Therefore, we judged the median number of 7 genes mutated per patient to be reasonable, since our selection of 555 genes was biased for genes known to be mutated in AML and we had a cut off at VAF of 20%.

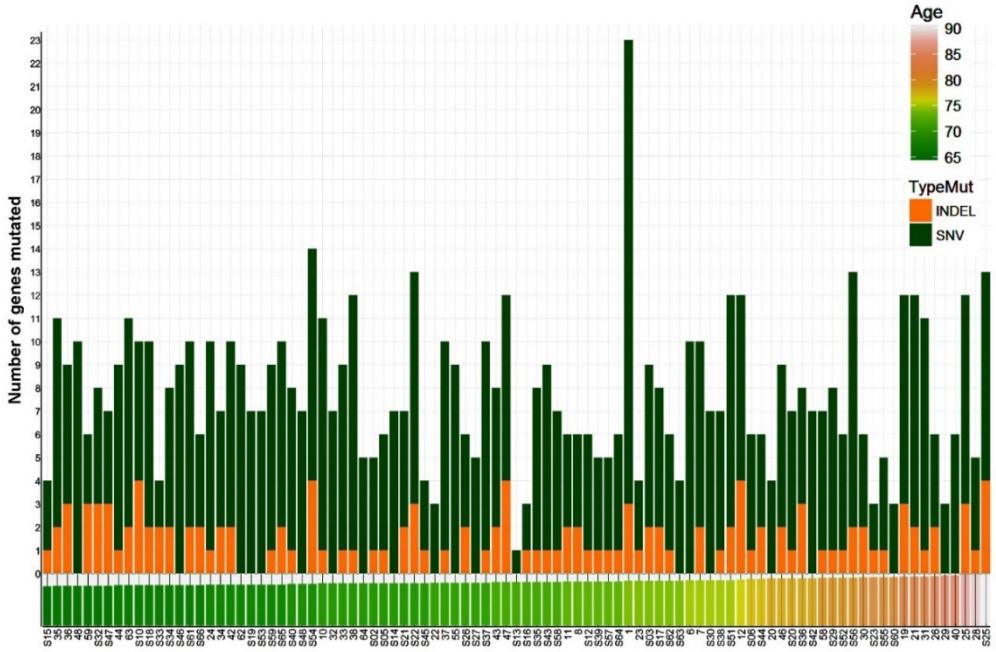


Figure 4.1.2 Frequencies of mutations in the SAL elderly AML for each patient.
The bar graph represents the number of genes mutated in each patient, organized by increasing patient age and annotated for the type of mutation found insertion or deletion (INDEL) or single nucleotide variation (SNV). The age of the patients at diagnosis is depicted by the heatmap code (in legend).

All patients had at least one mutation likely belonging to a major clone (in at least 37% of the tumor sample, as assessed by adjusted VAF), with exception of the patient with only 1 mutation (Figure 4.1.3A).

Furthermore, most of the altered genes displayed mutations of a possible founding clone, meaning adjusted VAF higher than the 37% (Figure 4.1.3B). The vast majority of the genes frequently mutated even showed a median adjusted VAF higher than the 37%. All of the genes with 3 or more mutations detected had at least one mutation belonging to a major clone, even if they seem to have a tendency for mutations with low VAF (for example *PDE4DIP*, see Figure 4.1.3B).

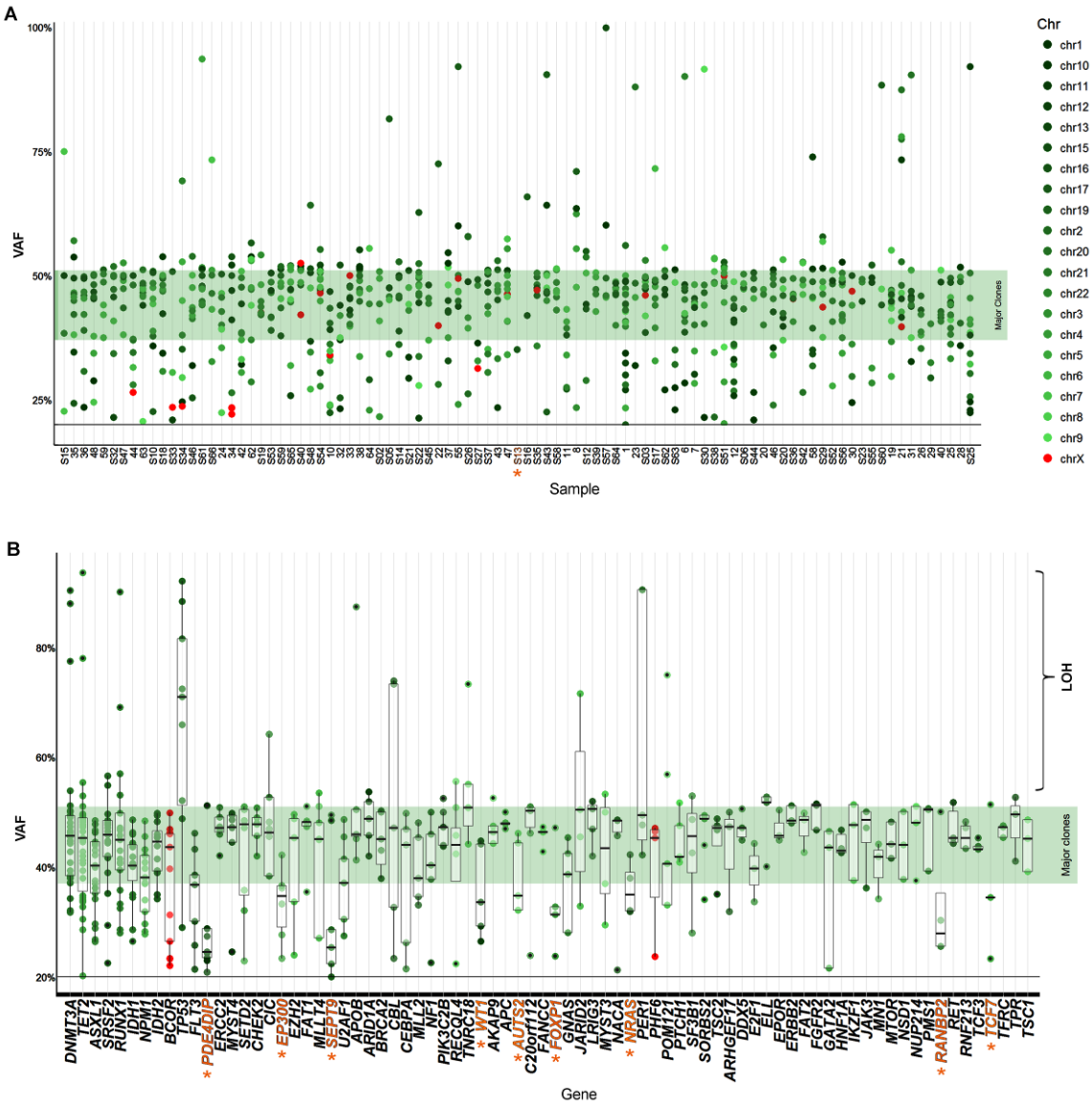


Figure 4.1.3 Variant allele frequencies of mutations in the SAL elderly AML.
Report of variant allele frequencies (VAF, adjusted for the number of X chromosomes) of mutations in SAL elderly AML, which are plotted: by sample (A, plotted by patient age at diagnosis 65 to 90 years) and by gene (B, where genes with at least 3 mutations). Green shades mark adjusted VAFs of mutations in a founding clone (37-51%). Mutations with higher adjusted VAFs were marked as loss of heterozygosity (LOH). A legend in the right panel lists the chromosome (Chr) where the gene is located. The orange asterisk denotes samples with only one mutation (A) or genes that had mutations with low adjusted VAFs (20-37%, B). Adapted from Silva *et al.* 2017[1].

We found no correlation between the numbers of mutated genes per sample and increasing age (either of Indel or SNV type, Figure 4.1.2), which was consistent with the observations within the elderly TCGA patient cohort (65 to 88 years old, Figure 4.1.4). An increase of mutation load with age was significant only when elderly (65 to 88 years old, n=60) were compared to younger patients (18 to 64 years old, n=137) within the TCGA cohort, (Figure 4.1.4). Despite the fact that these subgroups of

younger and older patients have different survival probabilities within the SAL cohort and within the TCGA cohort (Figure 4.1.5).

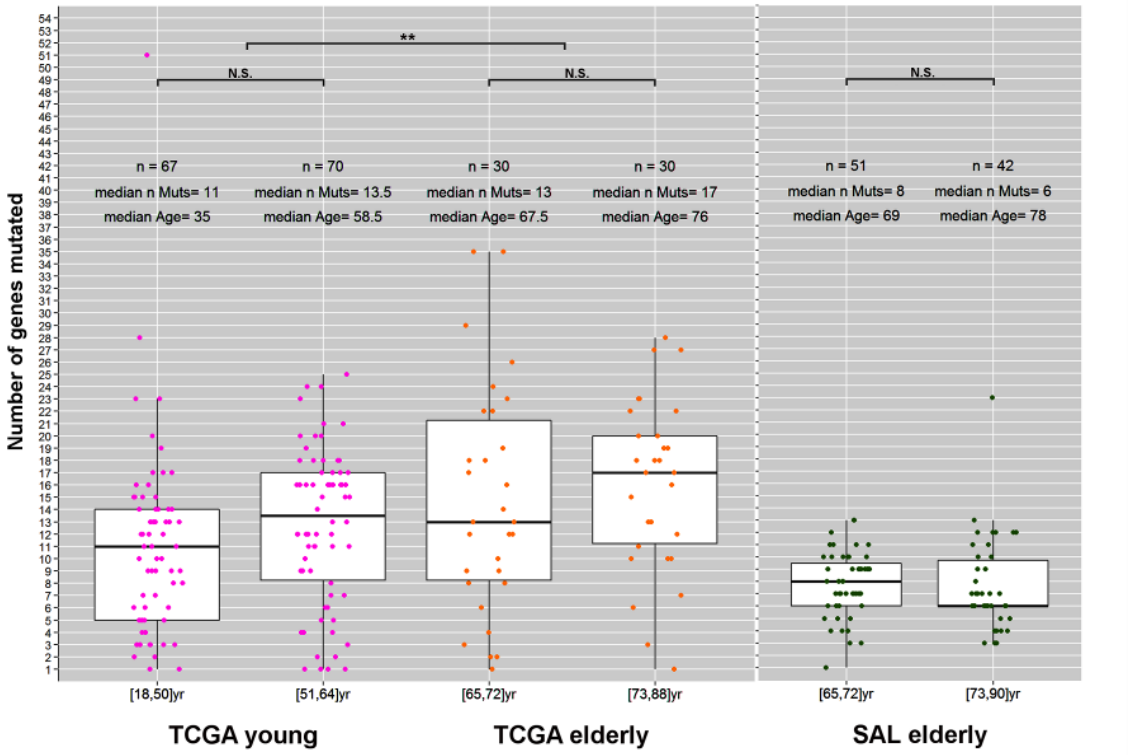


Figure 4.1.4 Rate of mutation of AML samples in the TCGA cohort and SAL cohort.

The boxplots show the number of genes mutated per sample found in TCGA cohort and in the SAL elderly cohort (including subdivisions of these into smaller age groups). The comparison depicts the increased number of genes with mutations from young patients (<65 years) to elderly (≥65 years), but not in further subdivisions of these TCGA groups (yr, years). The same was observed for the same subdivision of the SAL elderly cohort. Differences between groups were conducted with t-test analyses and are represented as N.S. non-significant and **p≤0.01. These were only accessed within each cohort due to the differences in the platforms of whole-exome of TCGA and our panel of exon sequencing.

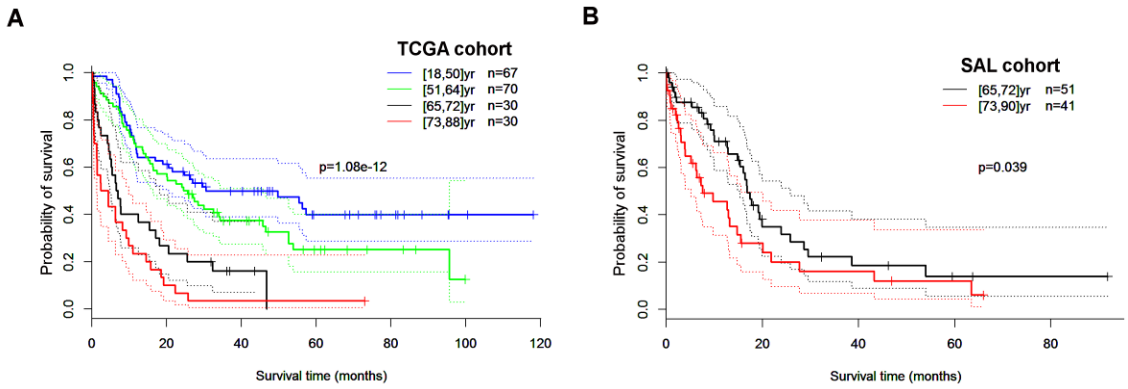


Figure 4.1.5 Survival analysis of AML patients in the TCGA cohort and in SAL cohort.

Kaplan-Meier curves show estimations of the overall survival of TCGA cohort (A) and of the SAL elderly cohort (B). The graphs show the differential outcome for the subdivisions of young (<65 years, yr) and elderly patients (≥65 years) into smaller age groups according to the median ages of 50y in young and 72y in elderly. Statistics were calculated with the log-rank test and the dotted lines mark the 95% confidence bounds of each curve.

In general, thirty-one genes were frequently mutated in the SAL elderly AML, as they were mutated in more than 5% of patients (Figure 4.1.6). The most frequently mutated genes were: *DNMT3A* 33.3%, *TET2* 25.8%, *SRSF2* 22.6%, *ASXL1* 21.5%, *RUNX1* and *IDH1* 17.2%, *NPM1* 16.1%, *IDH2* 10.8% (Figure 4.1.6).

Of note, we identified novel non-recurrent aberrations in previously reported cancer driver genes encoding proteins of the PI3K/mTOR pathway (PIK3CA, PIK3C2B, MTOR) and DNA damage proteins (BRCA2, ERCC2, FANCC).

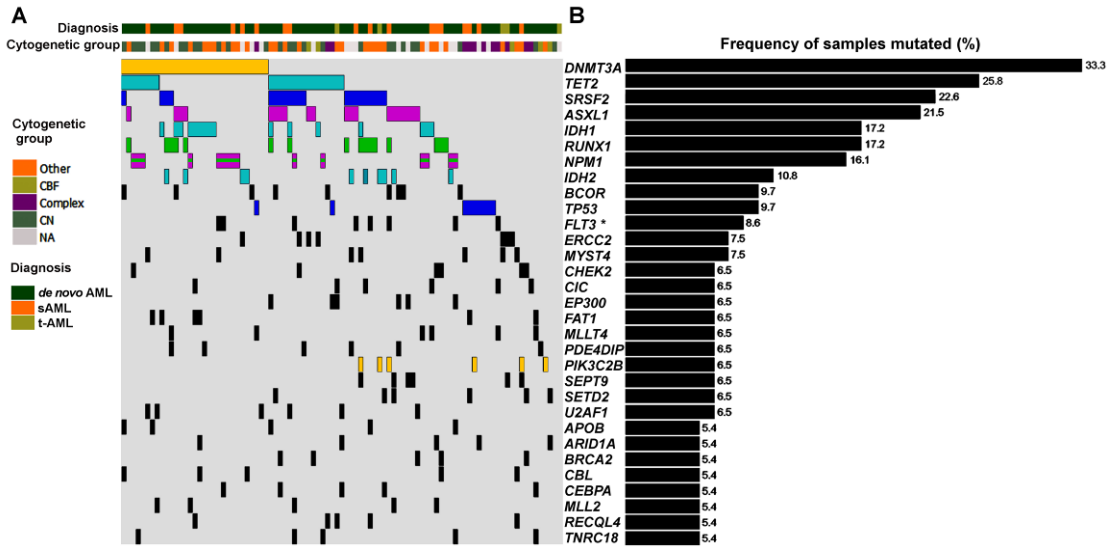


Figure 4.1.6 Recurrently mutated genes in the SAL elderly AML.
(A) Representation of mutations in genes (mutated in 5 or more samples) with patients represented in vertical and classified by cytogenetic groups. Classification of complex was defined for samples with 3 or more cytogenetic abnormalities (n=12, Complex), core binding factor included samples with t(8;21) or inv(16)/t(16;16) (n=5, CBF), samples with no cytogenetic abnormalities were designated cytogenetically normal (n=28, CN), others were samples with 1 or 2 cytogenetic abnormalities (n=30, other) and some were not available (n=18, NA). (B) The frequency of mutations in the genes was given in bars depicting the percentage of samples in the cohort with a mutation in the gene. Reporting NGS results only, not accounting for the *NPM1c* found only by Sanger sequencing and *FLT3* * refers to SNV not *FLT3*-ITD from the clinical data (in Table 3.1.1). Different colors mark pairs of genes with mutual exclusion: *NPM1* vs *ASXL1* (p=0.01, pink) and *NPM1* vs *RUNX1* (p=0.02, green), *IDH1* vs *IDH2* vs *TET2* (p=0.003, light blue), *DNMT3A* vs *PIK3C2B* (p=0.04, yellow), *SRSF2* vs *TP53* (p=0.04, blue). Adapted from Silva *et al.* 2017[1].

4.1.1 Frequently mutated epigenetic regulators

The frequency of mutations in epigenetic regulators was particularly high in the SAL elderly AML and affected 85% of patients (Figure 4.1.7). These mutations affected 30 genes that encode proteins belonging to several epigenetic protein families.

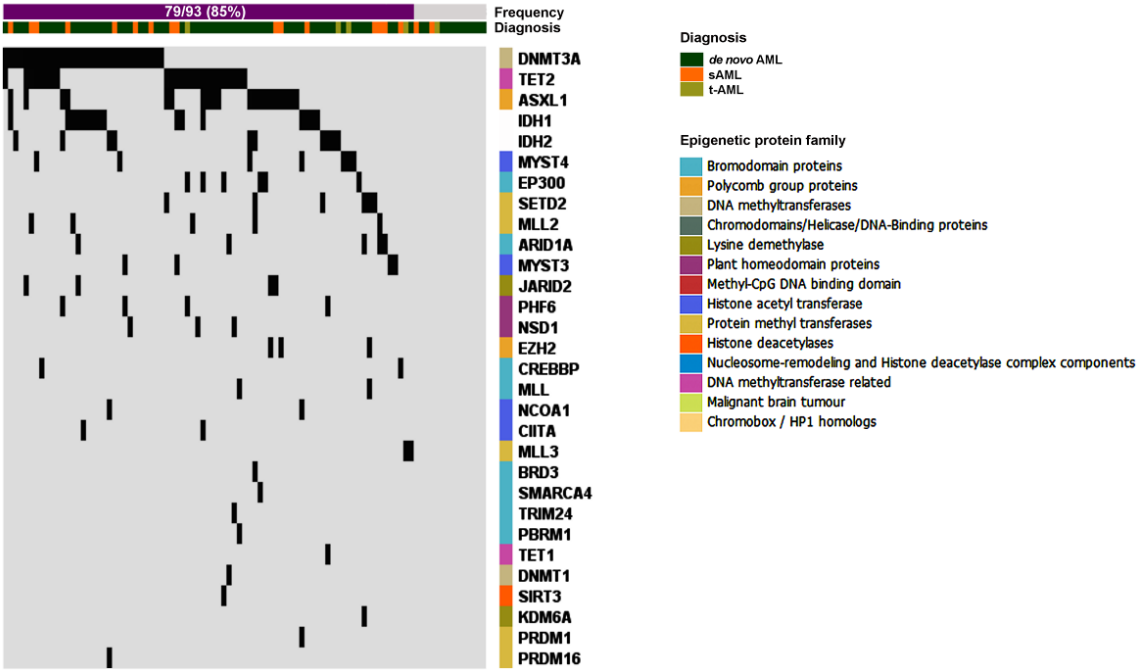


Figure 4.1.7 Mutations in epigenetic regulators in elderly AML.
Depiction of mutations found in epigenetic regulators with patients represented in vertical and genes classified into their protein families (sorted by co-exclusivity patterns). Patients were labeled with information collected at diagnosis as *de novo* AML (n=70), sAML (n=18) and t-AML (n=5).The frequency of patients with at least one of these genes mutated (in the bar) was calculated with the number of mutated/total number of patients (%). As presented in Silva *et al.* 2017[1].

Frequencies of mutations in *DNMT3A*, *TET2*, *ASXL1*, and *IDH1* were higher in the SAL elderly AML than previously reported for the general AML population[35,37,38,39,131]. In agreement with this result, in the TCGA young group (18 to 64 years old) we found 51% of samples with at least one mutation in an epigenetic regulator, while in the TCGA elderly group (65 to 88 years old) these mutations were present in 67% of samples (Figure 4.1.8).

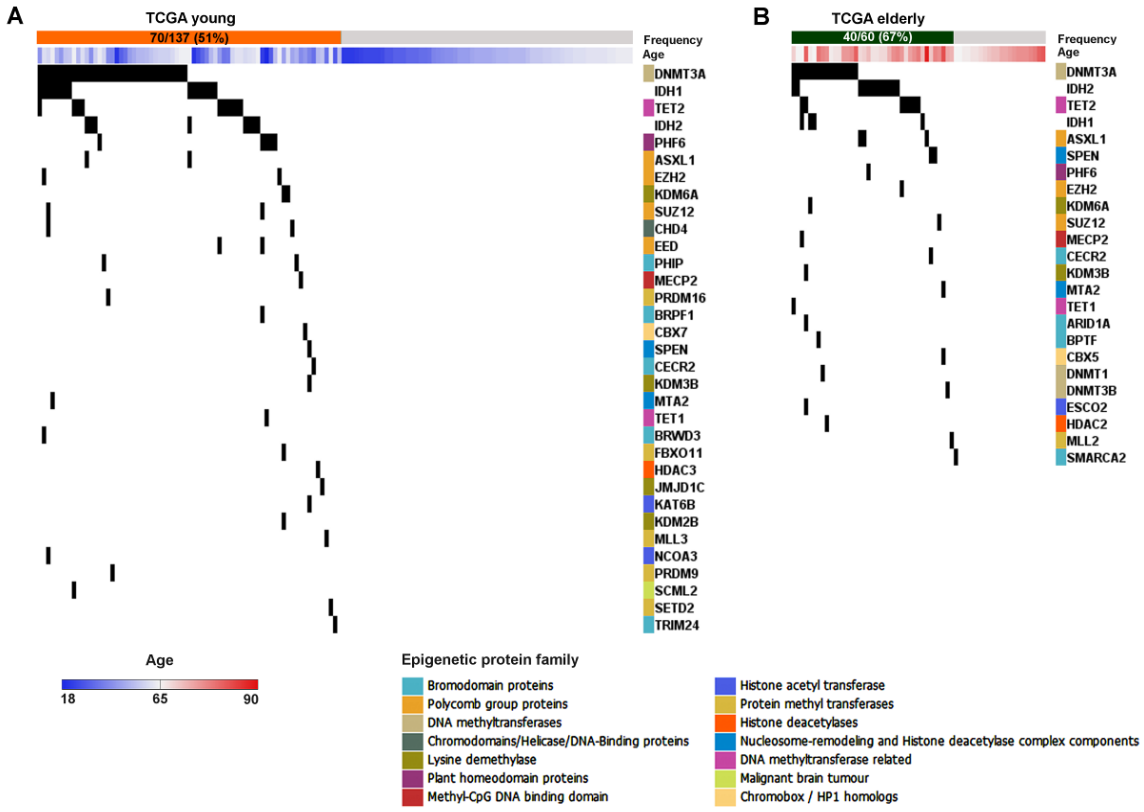


Figure 4.1.8 Epigenetic regulators mutated in TCGA elderly and TCGA young AML groups.

Representation of somatic mutations in 33 epigenetic regulators present in the TCGA young AML patients (A, n=137, <65 years) and 24 epigenetic regulators present in the TCGA elderly AML patients (B, n=60, patients ≥65 years). The frequency of patients with at least one of these genes mutated (in the bar) was calculated with the number of mutated/total number of patients (%). Genes were classified into their protein family and patients' ages were represented in heatmap code (in legend). As presented in Silva *et al.* 2017[1].

DNMT3A had the highest mutation frequency in our elderly AML cohort (33.3%) and has been previously reported to be mutated in 23-28% in other cohorts[37,39]. In addition, the incidence of *ASXL1* mutation with 21.5% (20/93) was much higher than the reported 3-6%[37,39,132]. A high rate of molecular alterations was observed for *IDH1/2* with a combined frequency of 28.0% (26/93). Of the 26 *IDH* mutations, 57.7% were *IDH1* R132 (15/26), 30.8% were *IDH2* R140 (8/26) and 8.3% were *IDH2* R172 (2/26). As expected molecular alterations in *IDH1* and *IDH2* were mutually exclusive and mutually exclusive with *TET2* (p=0.003, Figure 4.1.6). *TET2* mutations also showed a high mutation rate of 24% in elderly AML patients compared to the reported rate of 8-10% in other cohorts[37,39].

4.1.2 High rate of mutations in splicing proteins

We found frequent alterations in five genes of splicing factors including *SRSF2*, *U2AF1*, *SF3B1*, *ZRSR2*, and *DDX5*, affecting 38% of the AML elderly patients (Figure 4.1.9A). These are in a mutually exclusive manner (p=0.015), consistent with previous reports suggesting that these mutations elicit similar phenotypes[133,134]. Although most of these molecular alterations have been reported in MDS[133,135], in AML they have been reported only at low rates[133] (Figure 4.1.9B).

Strikingly, 22.6% of the SAL elderly AML patients (21/93) harbored *SRSF2* mutations (all alterations in residue P95), higher than the 8.3% or 10% recently reported in age unselected cohorts[38,39]. Thus, elderly AML had a distinct pattern of molecular alterations in splicing factors when compared to other AML entities such as *de novo* AML or sAML (Figure 4.1.9B), being present in patients with no clinical history of MDS.

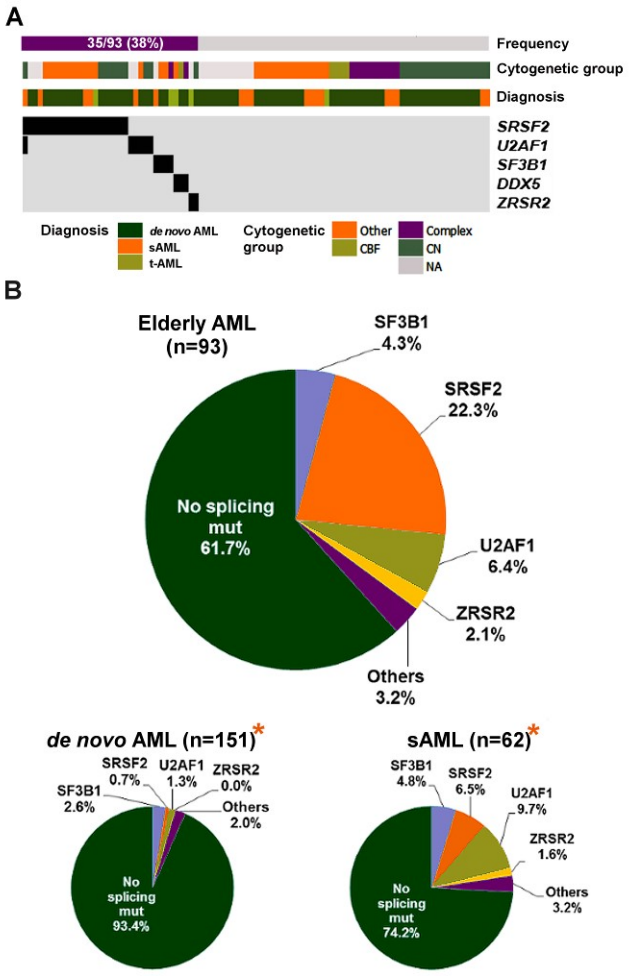


Figure 4.1.9 Frequency of mutations in splicing factors. (A) Illustration of mutations found in splicing factors with patients represented in vertical and genes in horizontal sorted by co-exclusivity (p=0.015). Patients were labeled with information collected at diagnosis as *de novo* AML (n=70), sAML (n=18) and t-AML (n=5). The cytogenetic groups were defined as: cytogenetically normal (CN), core binding factor (CBF), Complex (3 or more cytogenetic abnormalities), or other (1 or 2 cytogenetic abnormalities). The frequency of patients with at least one of these genes mutated (in the bar) was calculated with the number of mutated/total number of patients (%). Fisher exact tests (two-sided) were performed to compare cytogenetic and diagnostic characteristics of groups with and without mutations in splicing factors: no significant difference was observed for the characteristics. (B) Pie charts compare the percentage of alterations on splicing factors in elderly AML to other reported AML entities (*de novo* AML and sAML to a myelodysplastic syndrome, * data from Yoshida *et al.* 2011[133]). As presented in Silva *et al.* 2017[1].

The patients with these mutations did not present significant enrichments in particular patient diagnostic, cytogenetic or phenotypic characteristic that we could determine (Table 4.1.1).

Table 4.1.1 Description of the clinical characteristics of patients with mutations in splicing components.

The table reports the p-values from the Fisher tests (two-sided) performed for each characteristic between the two groups of patients (with and without mutations in splicing components). We classified patients using cytogenetic and diagnostic characteristic, as well as with the available data on the presence of at least one type of myelodysplastic features (megakaryocytic, granulocytic or erythroid dysplasia), which was termed as dysplasia, Dys. Patients were divided as: *de novo* AML, secondary AML (sAML), or therapy-related AML (t-AML). The cytogenetic groups were defined as: cytogenetically normal (CN, no detected cytogenetic abnormalities), core binding factor (CBF, includes t(8;21) or inv(16)/t(16;16)), Complex (3 or more cytogenetic abnormalities), or other (1 or 2 cytogenetic abnormalities).

Group	Mutations in splicing regulators	No mutations in splicing regulators	All	P-value
Diagnosis				
n	**35**	**58**	**93**	
***de novo* AML**, no. (%)	25 (71)	45 (78)	70 (75)	0.72
sAML, no. (%)	6 (17)	12 (21)	18 (19)	0.79
t-AML, no. (%)	4 (11)	1 (1.7)	5 (5)	0.06
Cytogenetic				
CN, no. (%)	10 (36)	18 (38)	28 (37)	0.90
CBF, no. (%)	1 (3.6)	4 (8.5)	5 (6.7)	0.64
Complex, no. (%)	2 (7.1)	10 (21)	12 (16)	0.19
Others, no. (%)	15 (54)	15 (32)	30 (40)	0.09
Dysplasia				
n	**17**	**32**	**49**	
Dys, no. (%)	10 (59)	19 (59)	29 (59)	1.00
No Dys, no. (%)	7 (41)	13 (41)	20 (41)	

4.2 Genetic alterations associated with elderly AML

Overall, several gene mutations appeared to have an age dependency in SAL elderly AML due to high or low frequencies when compared to AML reports for age unselected cohorts[37]. The associations between age and genetic alterations were made clear not only in our cohort but also in two other studies from Metzeler and colleagues[38] and Eisfeld and colleagues[34].

Therefore, we compared the frequency of mutations in the SAL elderly cohort (median age of patients was 72 years old) with the other 4 younger groups of patients from these 2 reports (Figure 4.2.1). Several tendencies for increased or decreased frequency of mutations with the groups' age became apparent in this analysis. Known mutations with strong correlations to WHO defined AML groups with adverse prognosis have higher frequencies with increasing age (*RUNX1*, *TP53* and *ASXL1*). In contrast, mutations that are correlated to WHO defined groups of favorable prognosis were less frequent in the older groups of patients (*NPM1* and *CEBPA*).

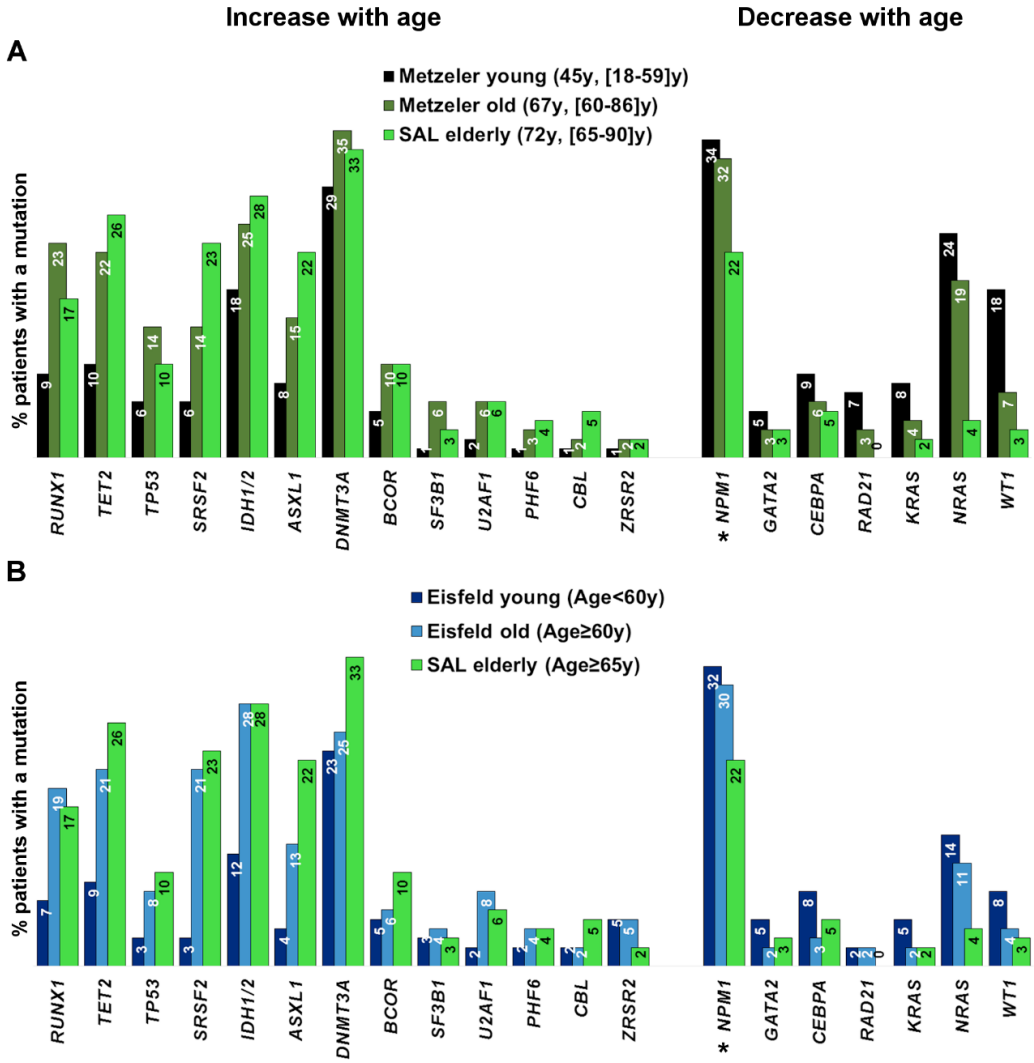


Figure 4.2.1 Mutation rates in the SAL elderly AML compared to mutation rates reported in two other cohorts.

Graphic representation of frequencies of mutations in genes, which have previously been associated with age. The frequencies obtained by gene panel approaches reported by (A) Metzeler *et al.*[38] (black and dark green, n=376 and n=288 respectively) and by (B) Eisfeld *et al.*[34] (dark and light blue, n=1080 and n=523 respectively) were compared to the obtained for the SAL cohort (light green, n=93). The legend shows the age restriction of each cohort with median and range of the ages in parenthesis when available. * In the SAL elderly AML *NPM1* rate is stated as the combined estimation of frequency of 22% (from correlation to Sanger results and retrieval from NGS detections with low coverage). Adapted in part from Silva *et al.* 2017[1].

4.3 Genetic heterogeneity in the SAL elderly AML

In the SAL elderly AML cohort, several combinations of genetic alterations originated genetically heterogeneous groups, similar to the ones found in age unselected AML cohorts.

Different mutational profiles were noticeable as the occurrence of several mutations does not appear to be random (Figure 4.1.6A). For instance, in spite of the general increase of mutations in epigenetic regulators and splicing factors, groups of patients with distinctive mutational patterns can be described. This could be seen by the co-exclusivity of patients with mutations in *IDH1* vs *IDH2* vs *TET2* (p=0.003) and *SRSF2* vs *TP53* (p=0.04). In the same way, in spite of the many *ASXL1* (21.5%) and *RUNX1* (17.2%) mutations, these were not present in patients with *NPM1* mutations. Furthermore, mutations in *TP53* were related to samples of complex karyotype, with 7 co-occurrences out of the 9 *TP53* mutated patients and out of the 12 complex karyotype samples (Pearson's correlation 0.63±0.08).

To further study genetic particularities of subgroups of elderly AML we obtained the survival data of several groups within the cohort. The expectation was that groups of patients with alterations in genes used to classify patients as adverse prognosis in other cohorts would also show poor survival in elderly AML. However poor prognosis within the elderly cohort could only be obtained for the group of patients with complex karyotype vs all others (log-rank p= 5.27×10^{-6}) or *TP53* mutated vs all others (log-rank p=1.2×10^{-3}). The other genes (and combinations) that have prognostic implications in other cohorts did not have differential survival probabilities in the SAL elderly AML. The mutations in *ASXL1* and *RUNX1* or even *DNMT3A*/*NPM1* combinations would be expected to be prognostic predictors in our cohort and were not. This was curious because the cohort had a relatively good median survival of 15 months (considering the patient's ages) and 79% of patients were treated with intensive chemotherapy regimens (Table 3.1.1).

On the other hand, the prognostic value of white blood cell count was still seen within the SAL elderly cohort. High WBC counts were indicators of poor prognosis for patients with values 35/nL or higher (Figure 4.3.1A). These groups with different WBC counts, the smoldering group (WBC<35/nL) or proliferative group (WBC≥35/nL) correlated to specific molecular marker contexts. Namely, *NPM1* and *FLT3* mutations (odds 0.069±0.001 and odds 0.23±0.06, respectively) were almost specific of the proliferating group, as *IDH1* (odds 11.3±1.6) mutated samples were of the smoldering group (Figure 4.3.1B).

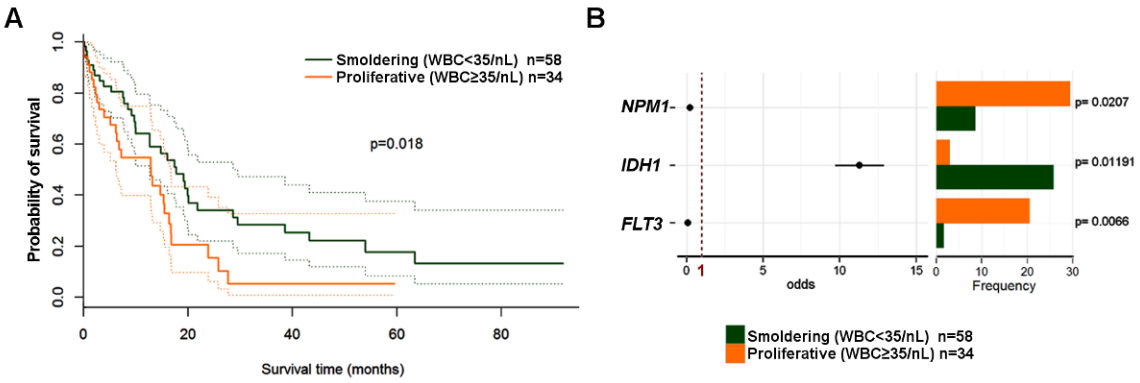


Figure 4.3.1 Mutations correlated to SAL elderly AML groups that were defined by WBC counts of prognostic relevance.

(A) Kaplan-Meier curves display the estimation of the overall survival for the division of the SAL elderly AML cohort into 2 groups, classified according to WBC counts at diagnosis into smoldering (lower than 35/nL) or proliferative (35/nL and higher). A significant difference between overall survivals was calculated with the log-rank test and dotted lines mark the 95% confidence bounds. (B) The odds ratio represents the skewness of mutation frequency in the SAL elderly groups smoldering versus proliferative (with odds 1 marked with dotted line). The corresponding frequency of mutations in each gene is plotted in horizontal bars, representing the % of mutated samples of each group. The p-values were calculated for the proportions of each gene in groups with Fisher exact tests corrected for the 281 genes mutated.

4.4 Protein networks and pathways of SAL elderly AML

To describe this complex molecular heterogeneity, we constructed a Reactome functional interaction (FI) network with the 139 genes that were mutated in more than one patient in the cohort. The network clustering algorithm identified highly interacting groups of proteins, establishing 8 different sub-networks (modules, Figure 4.4.1A). The four most prominent modules included 72 altered proteins and nearly every sample of the cohort (90 of 93). Module 1 was composed of NPM1 and mRNA processing proteins (RNA splicing and transport proteins), while Module 2 was composed of DNMT3A and DNA repair proteins. In turn, Module 3 was comprised of RUNX1, ASXL1 and other transcription factors (included prominent pathways NOTCH and WNT signaling), while Module 4 was formed by known oncogenes like FLT3 and KRAS/NRAS (mainly proteins with kinase activity, which included FGFR signaling molecules and the PI3K-AKT signaling pathway). Other modules were small and included BCOR, FAT proteins, and components of the mTOR signaling pathway.

4.4.1 Molecular patterns of mutations in mRNA processing and DNA repair proteins

Alterations in NPM1/mRNA processing (Module 1) and in DNMT3A/DNA repair proteins (Module 2) were spread across the cohort (p=0.004) and affected the majority of the patients 86% (80/93). Furthermore, alterations were mutually exclusive within each module: NPM1/mRNA processing (p=0.011) and DNMT3A/DNA repair proteins (p=0.052).

In addition, alterations in the two modules classified patients into four groups (Figure 4.4.1B): patients with mutations only in NPM1/mRNA processing proteins (Group A), patients with mutations in both the modules (Group B), patients with mutations only in DNMT3A/DNA repair proteins (Group C), and patients lacking any mutation in these modules (Group D). Patients of Group C (n=25) showed an inferior outcome, with a median OS of only 9 months, compared to patients with other mutation patterns (non-Group C, n=67, median OS 17 months, Figure 4.4.1C).

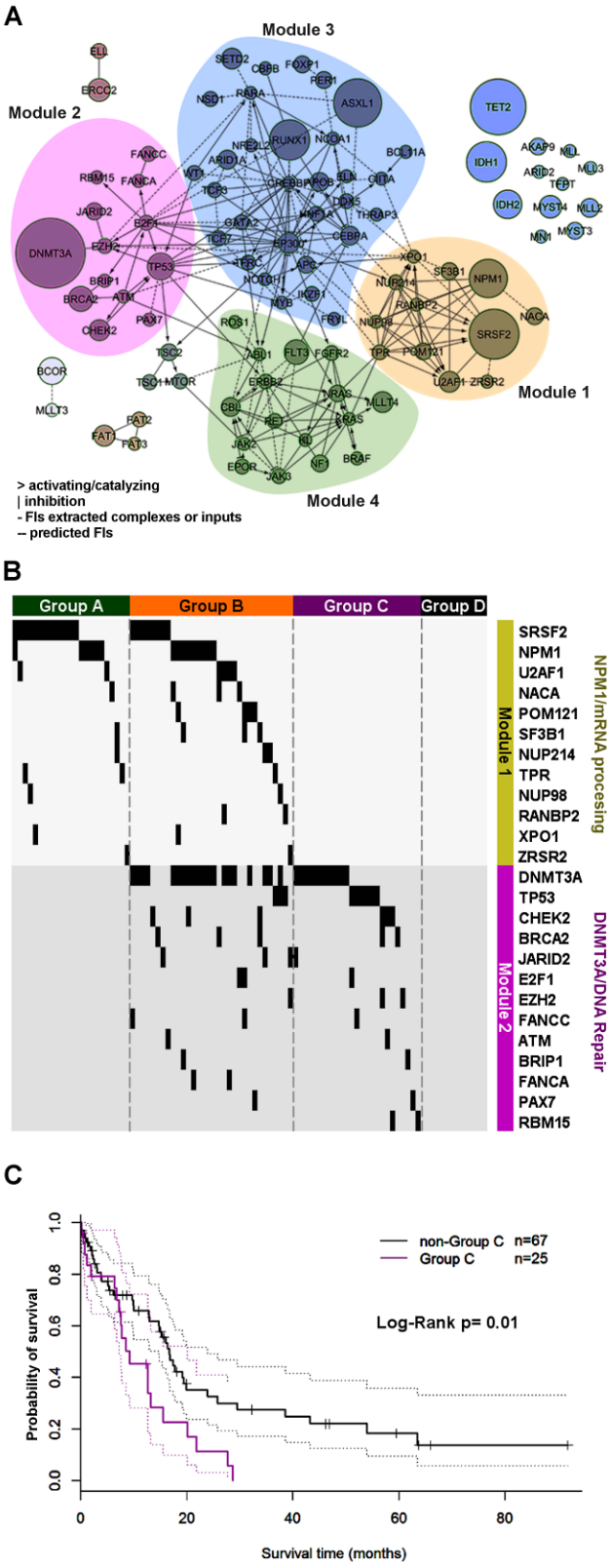


Figure 4.4.1 Reactome functional interaction network of altered proteins.

(A) Pathways affected by mutations were analysed with a FI network of proteins that clustered into 8 modules with 82 proteins. The 4 major modules were labeled as Module 1 (light green), Module 2 (pink), Module 3 (blue) and Module 4 (green). Some of the proteins that did not cluster in the FI network are also represented (light blue). The nature of the interactions is in the legend. (B) Representation of the division of the 93 AML into groups of samples with mutations in Module 1, in both Modules 1 and 2, only Module 2, or none of these modules (respectively Group A-D). (C) Kaplan-Meier estimation of the overall survival (n=92) showed the poor outcome of patients in Group C (log-rank test). Dotted lines mark the 95% confidence bounds. As presented in Silva *et al.* 2017[1].

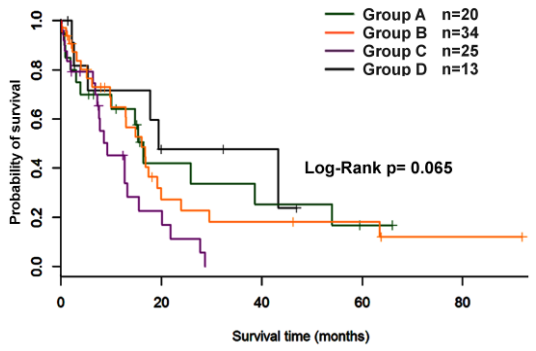
This was a tendency denoted in the survival curves of the four groups (Figure 4.4.2A). Apart from the genetic characteristics, Group C did not show enrichment of cytogenetic or clinical characteristics that could define it and be correlated to survival (Table 4.4.1).

Table 4.4.1 Characteristics of the groups defined by the Reactome functional interaction network analysis in the SAL elderly AML.

The table shows the clinical and molecular characteristics for the subgroups A, B, C, D within the Modules 1 and 2 (see Figure 4.4.1). The cytogenetic groups were defined as: cytogenetically normal (CN), core binding factor (CBF), complex (3 or more cytogenetic abnormalities), or other. Diagnosis were grouped by: *de novo* AML, secondary AML (sAML), or therapy-related AML (t-AML). Treatment regimens included standard intensive chemotherapy (induction treatment with 7+3 and high dose Ara-C consolidation), allogeneic stem cell transplantation (alloSCT), palliative (low dose Ara-C or hypomethylating agents) or best supportive care (BSC). The response was evaluated for patients receiving intensive induction chemotherapy, and relapses are shown for patients that had achieved a remission. Abbreviations used: white blood cell counts (WBC), Lactate dehydrogenase (LDH), bone marrow (BM). * The p-values were calculated by Fisher tests for the comparison of patients in Group C vs the remaining patients. As presented in Silva *et al.* 2017[1].

Group	A	B	C	D	All	P value*
Clinical characteristic						
n	**23**	**32**	**25**	**13**	**93**	
Age at diagnosis (y), median (ranges)	72 (66-86)	70 (65-90)	74 (66-88)	72 (69-80)	72 (65-90)	0.38
WBC (/nL), median (ranges)	26.2 (0.91-189.0)	14.9 (1.30-127.5)	10.2 (0.86-209.6)	14.7 (1.00-160.8)	14.8 (0.9-210)	0.56
LDH (U/L), median (ranges)	412 (138-3340)	402 (144-2015)	388 (161-4219)	516 (169-1482)	406 (138-4219)	0.97
BM Blasts, % (ranges)	82 (30-99)	74 (45-95)	74 (25-99)	72 (25-99)	75 (25-99)	0.19
Diagnosis						
***de novo* AML, no. (%)**	18 (78)	23 (72)	18 (72)	11 (85)	70 (75)	0.79
sAML, no. (%)	3 (13)	7 (22)	7 (28)	1 (8)	18 (19)	0.24
t-AML, no. (%)	2 (8.7)	2 (6.2)	0 (0)	1 (8)	5 (5)	0.32
Cytogenetic						
n	**16**	**26**	**21**	**11**	**75**	
CN, no. (%)	5 (31)	13 (50)	7 (33)	3 (27)	28 (37)	0.79
CBF, no. (%)	0 (0)	0 (0)	2 (9.5)	3 (27)	5 (6.7)	0.62
Complex, no. (%)	0 (0)	5 (19)	5 (24)	2 (18)	12 (16)	0.30
Others, no. (%)	11 (69)	8 (31)	7 (33)	4 (36)	30 (40)	0.67
Treatment						
n	**21**	**31**	**25**	**13**	**90**	
BSC or Palliative, no. (%)	4 (19)	7 (23)	7 (28)	1 (7.7)	19 (21)	
Intensive Chemotherapy	17 (81)	24 (77)	18 (72)	12 (92)	71 (79)	0.39
[no. alloSCT]	[2]	[6]	[1]	[2]	[11]	
Response to induction						
n	**17**	**23**	**18**	**12**	**70**	
Refractory, no. (%)	6 (35)	7 (30)	8 (44)	3 (25)	24 (34)	
Remission, no. (%)	11 (65)	16 (70)	10 (56)	9 (75)	46 (66)	0.39
[no. relapses]	[8]	[10]	[8]	[5]	[31]	0.46

A



B

Univariates	n/total	p-value	
Age (>72yr)	41/92	0.039	*
WBC (≥80/nL)	17/92	0.009	**
Complex Karyotype (vs others)	12/74	5.27E-06	***
Intensive Chemotherapie (vs others)	71/90	0.003	**
TP53 mt (vs wt)	9/92	0.001	**
Group C (vs non-Group C)	25/92	0.010	*

Multivariate	HR	SE (HR)	p-value	
Group C (vs non-Group C)	2.133	1.402	0.025	*
Age (>72yr)	1.097	1.357	0.763	N.S.
WBC (≥80/nL)	2.514	1.461	0.015	*
Complex Karyotype (vs others)	5.128	1.670	0.001	**
Intensive Chemotherapie (vs others)	0.152	1.695	3.56E-04	***

n= 75, number of events= 52 (18 observations deleted due to missingness)

Figure 4.4.2 Overall survival of the groups defined by the Reactome functional interaction network analysis in the SAL elderly AML.

(A) The Kaplan-Meier displays the estimation of the overall survival for the division of the SAL cohort into Group A, B, C and D using Modules 1 and 2 (see Figure 4.4.1; n=92). (B) The first table reports on all the factors with a significant effect on overall survival probability for the elderly AML cohort (univariates log-rank tests with p≤0.05). The second displays a multivariate analysis for all independent factors found for the SAL elderly AML (*TP53* mutations were not independent of complex karyotype). Hazard ratios (HR) were calculated with Cox's proportional hazard model. SE denotes standard error, N.S. is non-significant, *p≤0.05, **p≤0.01, ***p ≤ 0.001 and WBC denotes white blood cell count. As presented in Silva *et al.* 2017[1].

The prognostic significance of Group C was maintained when the analysis was restricted to patients that received intensive chemotherapy and in a multivariate analysis, the mutation profile of Group C was of independent adverse prognostic relevance (Figure 4.4.2B).

Importantly, using the TCGA cohort the prognostic significance of Group C could be confirmed (Figure 4.4.3), despite the difficulties of group division posed by the undetected *SRSF2* mutations[39] and some clinical differences of the cohorts (Table 4.4.2).

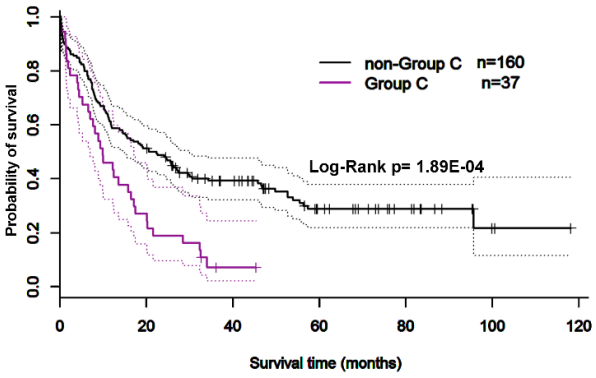


Figure 4.4.3 Overall survival of the groups defined by the Reactome functional interaction network analysis in the TCGA cohort.

Kaplan-Meier displays the estimation of the overall survival for the division of the TCGA cohort (n=197)[37] that showed the poor outcome of patients in Group C (log-rank test). Despite the age difference between our cohort (65 to 90 years) and the TCGA cohort (18 to 88 years) major clinical features and treatment characteristics were comparable. In both cohorts, the majority of patients were treated with standard intensive chemotherapy resulting in a similar median survival time. To note, more patients went on to alloSCT in the TCGA cohort, as these included a younger patient population. Dotted lines mark the 95% confidence bounds. As presented in Silva *et al.* 2017[1].

Table 4.4.2 Characteristics of SAL elderly AML cohort compared to the characteristics of the TCGA cohort.

Summary of the characteristics of patients: de novo AML, secondary AML (sAML), or therapy-related AML (t-AML). The cytogenetic groups were defined as: cytogenetically normal (CN), core binding factor (CBF, includes t(8;21) or inv(16)/t(16;16)), Complex (3 or more cytogenetic abnormalities), or other (1 or 2 cytogenetic abnormalities). Treatments included standard intensive chemotherapy or not (the latter consisted of palliative or best supportive care, which includes treatment with hypomethylating agents and a low dose of Ara-C). Several abbreviations were used: *FLT3* internal tandem duplications (*FLT3*-ITD), mutations in nucleophosmin altering tryptophan residues in the C-terminus of the protein (*NPM1c*), white blood cell counts (WBC), Lactate dehydrogenase (LDH), bone marrow (BM), allogeneic stem cell transplants (alloSCT), months (mon) and years (y).

	SAL cohort	TCGA cohort
Clinical characteristics	**n=93**	**n=200**
Age at diagnosis (y), median (ranges)	72 (65-90)	57 (18-88)
Sex (Male), no. (%)	47 (50.5)	108 (54)
***de novo* AML**, no. (%)	70 (75)	200 (100)
sAML, no. (%)	18 (19)	0 (0)
t-AML, no. (%)	5 (5)	0 (0)
LDH (U/L) , median (ranges)	406 (138-4219)	NA
WBC (/nL), median (ranges)	14.8 (0.9-210)	16.2 (0.4-298)
BM Blasts, % (ranges)	75 (25-99)	73 (30-100)
Cytogenetic	**n=75**	**n=195**
CN, no. (%)	28 (37)	92 (47)
CBF, no. (%)	5 (6.7)	12 (6.2)
Complex, no. (%)	12 (16)	24 (12)
Other, no. (%)	30 (40)	46 (24)
PML-RARA, no. (%)	0 (0)	18 (9.2)
BCR-ABL1, no. (%)	0 (0)	3 (1.5)
Molecular data		
***FLT3*-ITD**, no./ total no.(%)	15/70 (21)	NA
NPM1c, no./ total no.(%)	19/92 (21)	NA
Treatment	**n=90**	**n=200**
Palliative or Best Supportive Care, no. (%)	19 (21)	38 (19)
Intensive Chemotherapy, no. (%) [no. alloSCT]	71 (79) [11]	161 (81) [78]
Survival	**n=92**	**n=197**
Median survival time (mon)	15	17
Follow up time (mon), median (ranges)	7.4 (0.1-92)	47 (20-118)

4.5 Specific DNA methylation pattern of the elderly AML

First, we explored whether age and the high frequency of alterations in epigenetic regulators affected global DNA methylation. Thus, we combined the methylation data of the available TCGA cohort (n=194) and our SAL elderly AML cohort (n=79), performed unsupervised clustering and integrated somatic mutation information. In an unsupervised exploration, we found differential methylation in CpGs located in promoters that resulted in the separation of patients into two clusters with different age ranges (Figure 4.5.1). One cluster constituted mostly by elderly patients (n=118, median age of 70 years, ranging 27-90, 62% of the SAL cohort) and another cluster with younger patients (n=155, median age of 57 years, ranging 18-82, 38% of the SAL cohort).

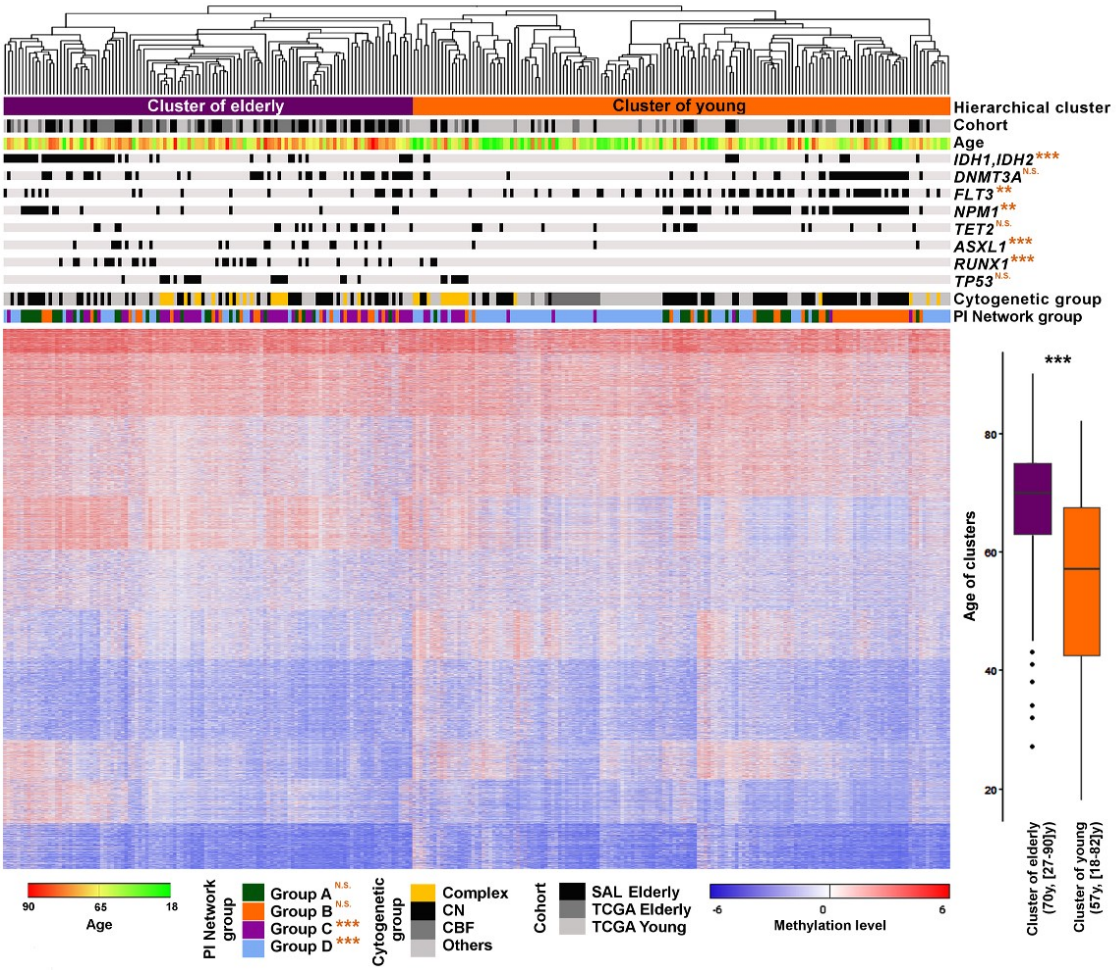


Figure 4.5.1 Methylation profiles of AML patients (TCGA cohort and SAL elderly).
The representation displays the methylation profiles of AML patients (TCGA and SAL cohorts combined), showing differential methylation patterns of elderly and young AML patients. (Caption continues in the next page)

Analysis of DNA methylation values in the 5,238 most variable CpGs in promoter regions (in the horizontal axis), scaled from unmethylated (−6, blue) to fully methylated (6, red). The depiction shows the unsupervised hierarchical clustering of 273 AML samples (in the vertical axis) with mutation information: 194 patients from the TCGA cohort (136 young of 18–64 years and 58 elderly of 65–88 years) and 79 patients from the SAL elderly cohort (65–90 years). Age of patients at the time of diagnosis is represented in the heatmap code (see legend). Samples considered part of the two hierarchical clusters were labeled as cluster of the elderly (n=118, purple) and cluster of the young (n=155, orange), due to the significantly different median age of the patients (70 years and 57 years, respectively, ***P≤0.001 obtained by t-test, boxplot in the right, median and range of ages are annotated). Patients were classified in cytogenetic groups: cytogenetically normal (CN), core-binding factor (CBF), complex (3 or more cytogenetic abnormalities) or others. *FLT3* included both SNVs and *FLT3*-ITDs available and *NPM1* included both mutations identified by NGS and Sanger sequencing. Adjusted Fisher exact tests for skewness of mutations between the cluster of the elderly and cluster of the young is specified on the right of each gene, or of the legend in case of the groups found using the protein functional interaction network (PI network, NS denotes non-significant, *P≤ 0.05, **P≤0.01 and ***P≤ 0.001). Adapted from Silva *et al.* 2017[1].

Of note, these two epigenetic clusters were markedly different in genetic alterations (Figure 4.5.1), as most of the samples with mutations in *IDH1/2* (79%), *ASXL1* (86%), *RUNX1* (90%), *TP53* (72%) and co-mutated for *IDH/DNMT3A* (73%) were in the cluster of the elderly. In contrast, the cluster of young patients was enriched for *DNMT3A*/*NPM1*/*FLT3* triple mutated samples (88% of these triple mutated samples were in this cluster).

Comparing the cluster of the elderly with the cluster of the young, we identified many differentially methylated regions (n=1,641), while using categorical age groups elderly (TCGA+SAL, ≥65 years) vs young (TCGA, <65 years) 4.6x fewer regions were identified (n=356, Figure 4.5.2A). Hypomethylated regions in the cluster of the elderly (hypoMRs, n=662) were almost all in genes (83%) and more than half overlapped promoters, while the hypermethylated regions (hyperMRs, n=979) although mostly overlapping genes were less associated to promoter regions or transcription start sites (Figure 4.5.2B, complete lists of regions are in Appendix C).

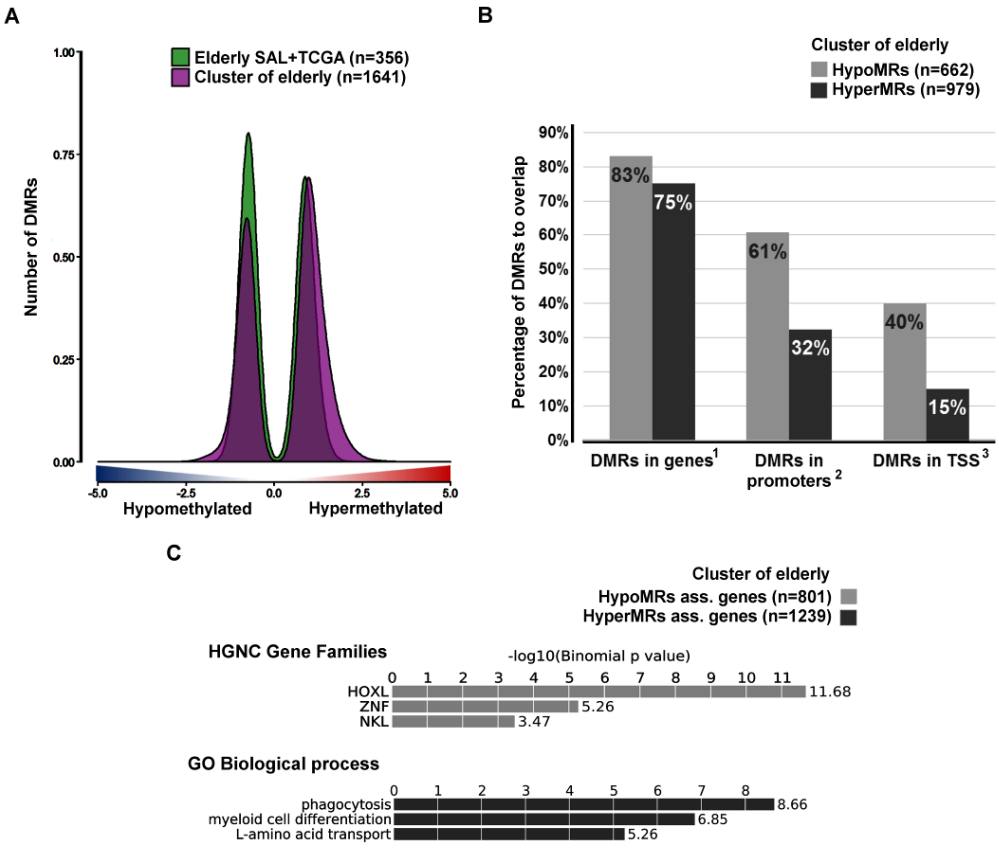


Figure 4.5.2 Differential methylation patterns of the elderly in comparison to young.

(A) The density graph reports the significant differential methylated regions (DMRs) between the first level hierarchical clusters from the unsupervised analysis (cluster of elderly vs cluster of young), or between the chronological age groups (elderly TCGA and SAL ≥65y vs young TCGA <65years). These regions were obtained from the 104,675 most variant CpGs (n is the total number of DMRs). (B) The bar graph shows the percentage of DMRs between the hierarchical clusters associated with specific gene regulatory features. DMRs were classified into hypermethylated (hyperMRs, *n*=979) or hypomethylated (hypoMRs, *n*=662) according to their ratio of methylation in the cluster of elderly/cluster of young (n is the total number of DMRs hypomethylated and hypermethylated). The associations of DMRs to 3 gene regulatory features were examined: [1]DMRs with at least 1b overlapping a gene; [2]DMRs within 1kb of a gene promoter; [3]DMRs with at least 1b overlapping to a transcription start site (TSS). Associations calculated in EpiExplorer[128]. (C) The graph is a representation of enrichments in gene families and biological processes of genes associated (ass.) to the DMRs, for the genes associated to hyperMRs (n=1,239) and for the genes associated to hypoMRs (n=801) connected to the cluster of the elderly. Associations to genes and enrichments were calculated by GREAT[125]. Adapted from Silva *et al.* 2017[1].

Hypomethylated regions (n=662) in the cluster of the elderly were associated with genes (n=801) responsible for morphogenesis (HOXL and NKL gene families), while hypermethylated regions (n=979) were related to genes (n=1,239) associated with phagocytosis, myeloid differentiation and amino acid transport (Figure 4.5.2C and detailed in Figure 4.5.3).

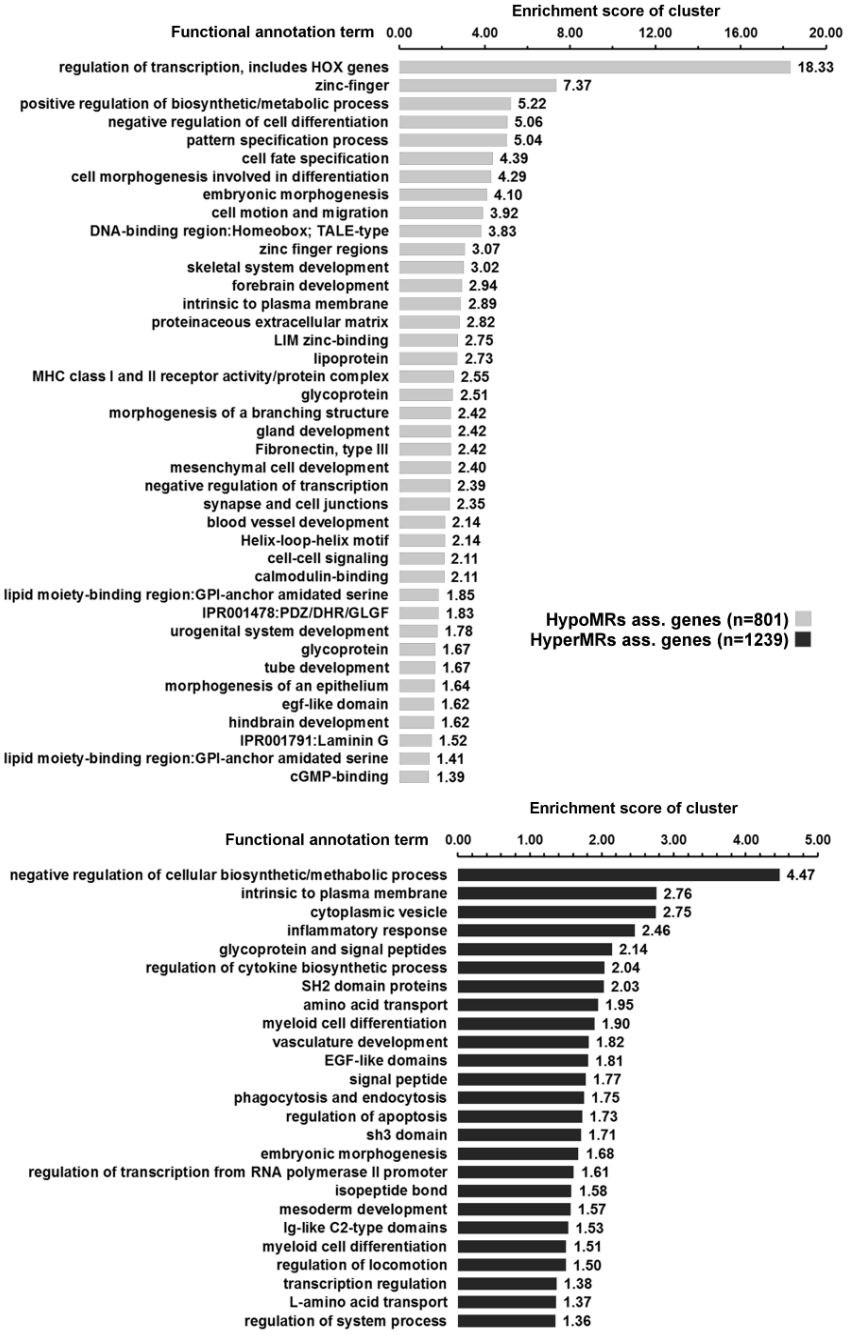


Figure 4.5.3 Enrichment of biological terms for genes associated with the differential methylation signature in the elderly.

Biological terms enriched in the list of genes associated with the DMRs from the comparison of the first level hierarchical clusters of the unsupervised analysis (cluster of elderly vs cluster of young). Regions were classified as hypermethylated (hyperMRs) or hypomethylated (hypoMRs) in relation to the cluster of the elderly AML. DMRs' associations to genes were calculated in GREAT[125] (n was the number of ass. genes). Enrichment scores of each functional annotation cluster were calculated using DAVID (EASE scores with FDR<10%)[126,127]. As presented in Silva *et al.* 2017[1].

4.5.1 Genomic locations of elderly AML differential methylation

We were attentive to the importance of defining genomic regions where the methylation levels are unique. Large epigenetic changes that occur in specific regions of the genome might be of additional significance.

So, we analysed particular patterns in the distribution of the elderly AML DMRs across genomic positions. We made two main observations: 1) most chromosomes had comparable amounts of hyperMRs and hypoMRs, except for chromosomes 16 and 17 where the number of hypermethylations in samples of cluster of elderly AML was clearly high (Figure 4.5.4A) and 2) there was a tendency of the hyperMRs to be located in the tips of chromosomes (Figure 4.5.4B).

The higher methylation levels of the samples from the cluster of the elderly were more striking in chromosome 17. This was the chromosome with the highest number of hyperMRs (109 of the 979, 11%) and many of these regions were located in the q25.3 band (Figure 4.5.4B, box). This was an unusual concentration of hypermethylation on the tip of chr17 in which 35 hyperMRs (35/109, 32%, Table 4.5.1) were concentrated in about 2.4Mb (highest density area in the blue box of Figure 4.5.4B).

Within this area, there were 2 major regions strongly hypermethylated, the regions related to neighboring genes *ACTG1/KIAA1447* and to *RPTOR*/*CHMP6*. These were the 4 genes that had the highest number of hypermethylated regions from the cluster of elderly associated to them. There were 9 hyperMRs per each pair of genes (*ACTG1/KIAA1447* and *RPTOR*/*CHMP6*) and each of these regions related at least to 3CpGs.

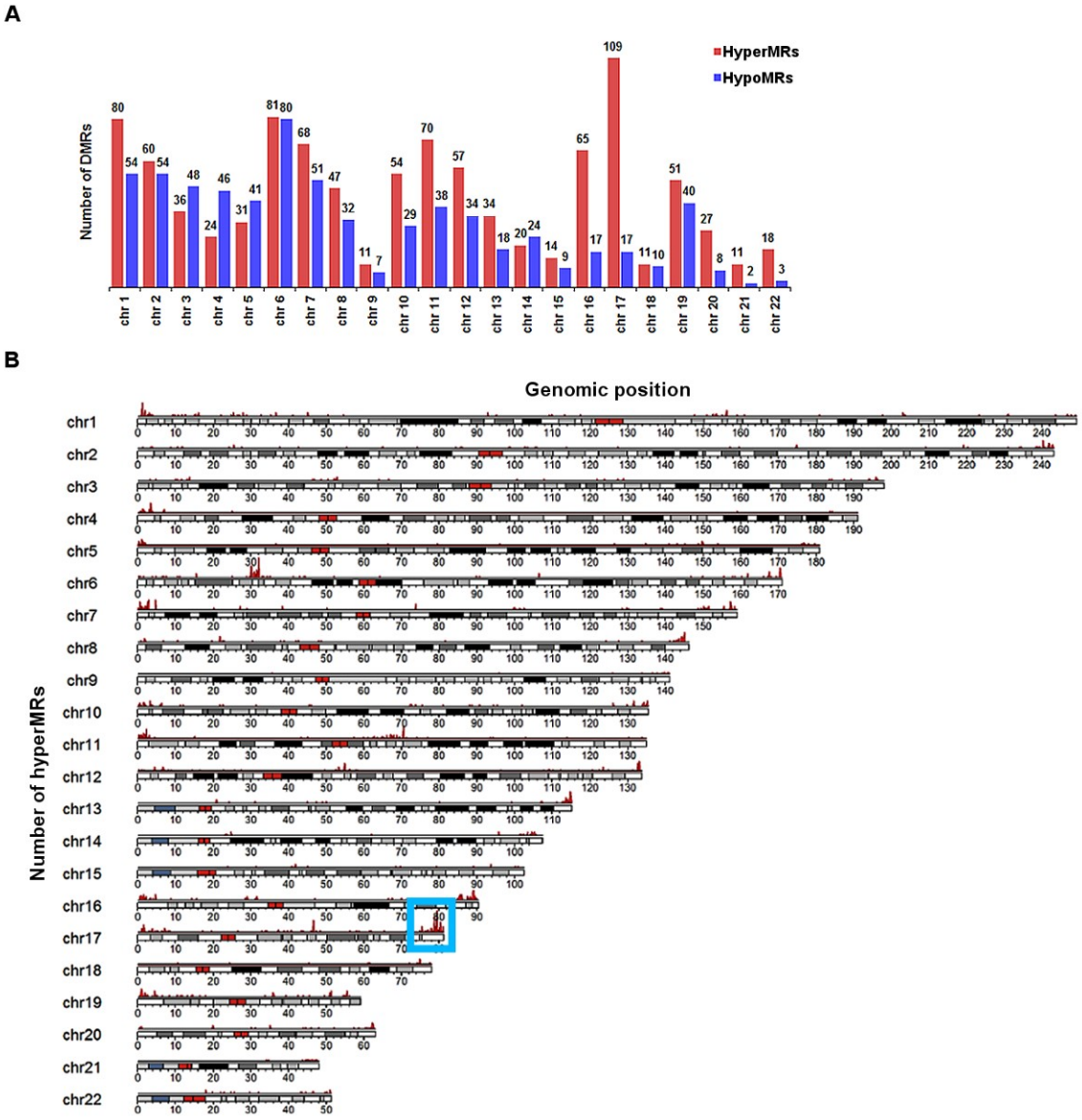


Figure 4.5.4 Distribution of DMRs of the cluster of elderly AML across chromosomes.

(A) The bar graph reports the number of DMRs from the cluster of elderly vs cluster of young in each chromosome (comparison of hierarchical clusters in Figure 4.5.1). These were classified as hypermethylated (hyperMRs, in red) or hypomethylated (hypoMRs, in blue) according to the methylation level in the cluster of elderly AML. (B) The graph shows the hyperMRs plotted according to their genomic position from the start of the short arm to the end of the long arm (oriented left to right) of chromosomes 1 to 22. The density plots represent the number of hyperMRs calculated by partitioning the genome in 0.2Mb regions. Chromosome 17q25.3 band was highlighted with a blue box. The scale of genomic regions is given in Mb. Red region of chromosome ideograms marks the centromeres.

Table 4.5.1 HyperMRs of the cluster of elderly AML concentrated in chr17 q25.3.
The table shows genomic positions of 35 regions obtained with the bumphunter algorithm as hypermethylated in the cluster of elderly (Figure 4.5.1) and the genes correlated to each region. These are spanning the region chr17:77901030-80292252 (using assembly hg19). Each gene association was calculated by GREAT[125] with the predefined association rule: Basal + extension: 5kb upstream, 1kb downstream, 1Mb maximum extension and curated regulatory domains included. The distance of each gene to the region CpGs is given in parentheses.

Chr	Start	End	Genes correlated (distance to gene in bases)
chr17	77901030	77901317	CBX4 (-87946), TBC1D16 (+108473)
chr17	78058704	78058778	GAA (-16762), CCDC40 (+48306)
chr17	78190755	78190983	SLC26A11 (-3360), SGSH (+3330)
chr17	78560478	78560916	CHMP6 (-404944), RPTOR (+41629)
chr17	78735268	78735596	CHMP6 (-230209), RPTOR (+216364)
chr17	78748077	78748494	CHMP6 (-217355), RPTOR (+229218)
chr17	78753273	78754372	CHMP6 (-211818), RPTOR (+234755)
chr17	78755379	78755442	CHMP6 (-210230), RPTOR (+236343)
chr17	78829626	78829930	CHMP6 (-135863), RPTOR (+310710)
chr17	78851149	78851262	CHMP6 (-114435), RPTOR (+332138)
chr17	78854232	78854418	CHMP6 (-111316), RPTOR (+335257)
chr17	78865263	78866235	CHMP6 (-99892), RPTOR (+346681)
chr17	79004850	79006166	BAIAP2 (-3454)
chr17	79109729	79109910	AATK (+29997), BAIAP2 (+100858)
chr17	79128918	79129078	AATK (+10819), BAIAP2 (+120036)
chr17	79228937	79229385	C17orf89 (+16122), SLC38A10 (+39944)
chr17	79282893	79283959	SLC38A10 (-14321), TMEM105 (+21048)
chr17	79297435	79297618	SLC38A10 (-28422), TMEM105 (+6947)
chr17	79374327	79374741	KIAA1447 (+994)
chr17	79377272	79378207	KIAA1447 (+4200), ACTG1 (+102067)
chr17	79380400	79380706	KIAA1447 (+7013), ACTG1 (+99254)
chr17	79419796	79420424	KIAA1447 (+46570), ACTG1 (+59697)
chr17	79425877	79426432	KIAA1447 (+52615), ACTG1 (+53652)
chr17	79428036	79428750	ACTG1 (+51414), KIAA1447 (+54853)
chr17	79429330	79429729	ACTG1 (+50277), KIAA1447 (+55990)
chr17	79432709	79433150	ACTG1 (+46877), KIAA1447 (+59390)
chr17	79442037	79442206	ACTG1 (+37685), KIAA1447 (+68582)
chr17	79484960	79485709	FSCN2 (-10223), ACTG1 (-5528)
chr17	79799358	79799549	PPP1R27 (-6545), P4HB (+19116)
chr17	80159399	80159595	FASN (-103289), SLC16A3 (-32066)
chr17	80190129	80190154	SLC16A3 (-1421)
chr17	80196719	80197538	SLC16A3 (+5566), CSNK1D (+34478)
chr17	80266884	80267030	CSNK1D (-35350), CD7 (+8521)
chr17	80273242	80273685	CSNK1D (-41857), CD7 (+2014)
chr17	80292079	80292252	SECTM1 (-292)

When examined in detail different CpGs across this 2.4Mb region have different factors that correlate with their methylation levels. Some CpGs seem strongly affected by mutations and other clearly more related to patients' age. As an example of this, we show the regions of the 3 adjacent genes *KIAA1447*, *ACTG1* and *FSCN2* (Figure 4.5.5). Some CpGs were clearly different between the 2 hierarchical clusters (examples in boxes A and B), while others were particularly hypomethylated in *DNMT3A/NPM1/FLT3* mutated samples (example in box C) and/or very strongly hypermethylated in the *IDH1/2* mutated patients (example in box D).

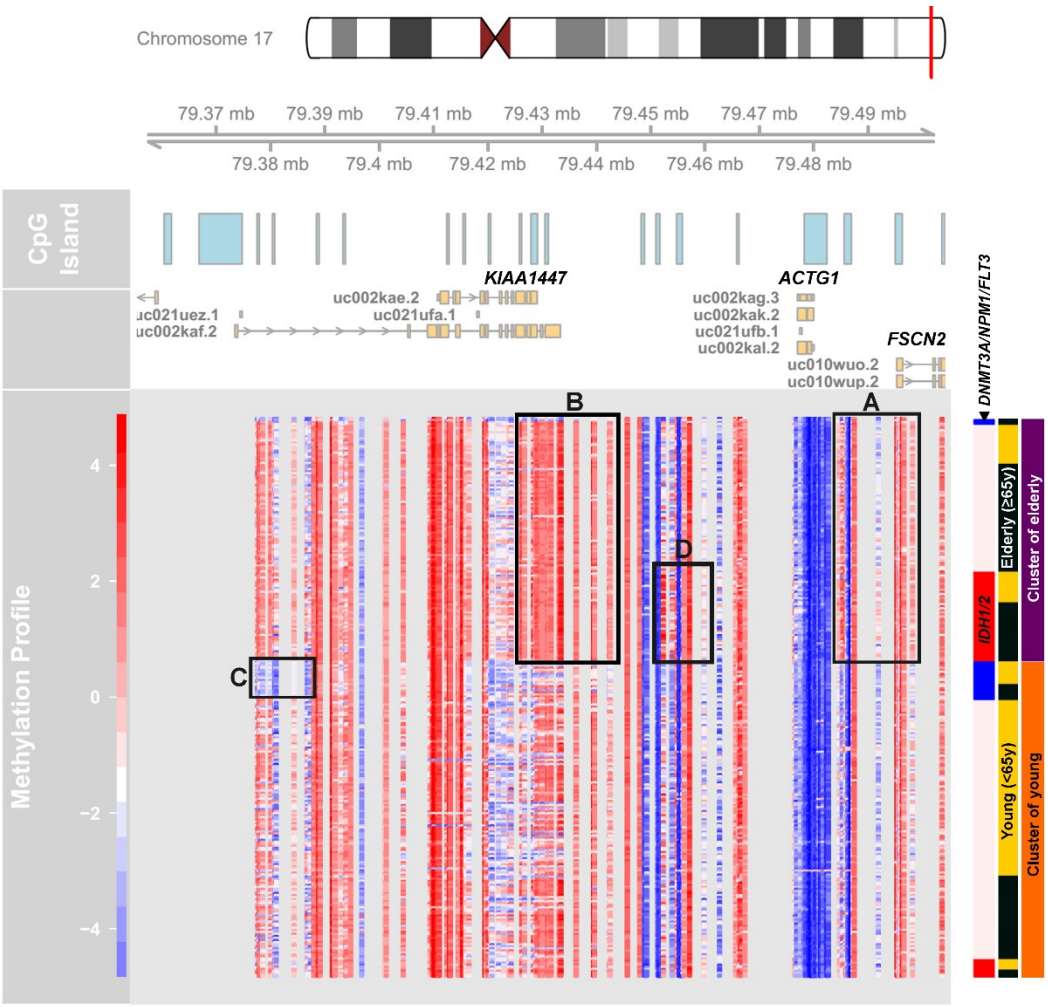


Figure 4.5.5 Distinct methylation profiles of elderly in the *KIAA1447/ACTG1/FSCN2* region.

DNA-methylation from the CpGs in the tip of the long arm of chromosome 17 using beta-values scaled from unmethylated (-5, blue) to fully methylated (5, red). In the depiction the 273 AML samples from the two cohorts (TCGA and SAL) are in horizontal and positions of CpGs according to GRCh37/hg19 are in vertical. The CpG islands of the genomic region are marked with blue boxes. Patients are annotated according to the group of the 1st division of hierarchical clustering (cluster of elderly, purple annotation, or young, orange annotation, Figure 4.5.1), age group according to patient chronological age (elderly for ≥65 years old, black annotated, or young for <65 years old, yellow annotation) and mutation information for 2 relevant high rate mutated groups (*IDH1/2*, red annotation, and triple mutated *DNMT3A/NPM1/FLT3*, blue annotation).

The particular hypermethylation pattern of the tip of chr17 overlapped several genes that were part of endocytosis and phagocytosis processes (see gene enrichment in Figure 4.5.3), with emphasis on functions in actin organization. The *ACTG1* codes for the gama-actin in the cellular cytoskeletons and is one of the genes in the biological process enrichment phagocytosis of the hyperMRs[136]. Gama-actin is also important for cell adhesion, being involved with the cadherins/protocadherins and catenins for the regulation of adherens junctions[136] (families of proteins for which we

found many other DMRs). The third gene *FSCN2* (Fascin Actin-Bundling Protein 2, Retinal) codes a protein with an actin-binding function which is expressed only in retina and blood[136].

Interestingly, in their study Bullinger and colleagues in 2010[137] connected the hypermethylation in this region of the chr17 long arm overlapping the neighboring genes *KIAA1447* and *ACTG1* to poor survival in AML (these confirmations are marked with * in Table 4.5.2B, next chapter). Not much is known about *KIAA1447,* an alias of *BAHCC1* (BAH Domain And Coiled-Coil Containing 1). This is a gene encoding a protein with a chromatin binding domain, interacting with the chromatin silencing complex for transcription regulation[136].

Furthermore, *BAIAP2* is the gene that encodes BAI1 Associated Protein 2, a protein that functions as an insulin receptor tyrosine kinase substrate in the brain[136]. It interacts with CDC42, being necessary for the CDC42-mediated reorganization of the actin cytoskeleton and whose RNAi phenotype is a reduction of endocytosis. Moreover, *CHMP6* is also involved in endocytosis and is in this genomic region[136]. CHMP6 is a probable core component of the endosomal sorting required for transport complex III which is involved in multivesicular bodies formation and sorting of endosomal cargo proteins[136].

Besides *BAIAP2*, in this region were two other genes of proteins involved in metabolism, namely in insulin response, *RPTOR* and *FASN*. The gene *RPTOR* codes for the protein Regulatory Associated Protein Of MTOR Complex 1[136]. RPTOR or RAPTOR is involved in the control of the mammalian target of rapamycin complex 1 (mTORC1) activity which regulates cell growth, survival, and autophagy/longevity in response to nutrient and hormonal signals[136]. A DMR overlapping *RPTOR* was found in the most recent paper reporting DMRs with correlations to survival probability (using this TCGA methylation data)[138]. They advanced this gene to have one of the 8 DMRs signature that could be used as a prognostic marker and have a crucial role in AML.

In turn, *FASN* codes for the Fatty acid synthase, which main function is to catalyze the synthesis of palmitate from acetyl-CoA and malonyl-CoA and is a downstream responder in the insulin signaling pathway[136].

4.5.2 Differentially methylated genes in elderly AML

The gene expression of particular genes has proved to be very relevant for age phenotypes, for cancer development and more specifically for AML. Therefore, we wanted to report the levels of DNA-methylation that could affect specific genes known to be involved in these phenotypes.

Additionally, we looked at the DMRs found for the cluster of elderly to identify regions/genes with DNA-methylation levels that were previously correlated to survival in AML. These genes could constitute candidates for further use as predictors of poor survival of AML patients. We also wanted to highlight which of these genes could have expression levels correlated to survival in AML. To access this we obtained the Kaplan-Meier curves of the patients with low versus high gene expression, using the RNA-seq data of the TCGA cohort. Hence, we examined the survival of the groups of patients with lower than mean expression value vs the group of higher than mean expression value for the individual genes of interest.

Our results from the application of these methods are reported in Table 4.5.2 A-C, showing the genes we identified as differentially methylated in the cluster of elderly AML in our dataset (n=2040) that are also known to be involved in specific phenotypes.

Table 4.5.2 Genes in DMRs of the cluster of elderly AML which are related to age, cancer and AML pathogenesis or survival.

The tables show the genes correlated to regions of hypo and hypermethylation in the cluster of the elderly that are involved in age and cancer (A) or AML (B and C). (A) The genes overlapping age-related genes[139] and oncogenes or tumor suppressor genes (TSG) according to the 20/20 rule[140]. (B) The genes overlapping genes that are correlated to DMR signatures of AML samples (CD34+_vs_AML[88]: AML compared with normal bone marrow CD34+ cells and Ctr_vs_AML[137]: AML compared to blood from healthy donors). (C) The genes overlapping genes correlated to DNA methylation regions with prognosis value previously reported in 3 papers: Marcucci *et al.*[95], Li *et al.*[94] and Figueroa *et al.*[89]. Bold marks the genes for which the cluster of elderly showed both hypo and hypermethylated regions. Blue marks genes that were reported in the respective study to be hyper and hypomethylated depending on the AML group that was compared and not in the overall AML. Green and red mark the genes we could confirm to be of prognosis value in the TCGA samples with the appropriate correlation to the DNA methylation alteration. Green marks the bad prognosis for the low expression of the gene, while red marks the bad prognosis for the high expression of the gene. The * marks genes that are part of a big region of hypermethylation on chr17 shown in Figure 4.5.5.

A

hypoMRs	hyperMRs
Age Genes (21/307)	
CTF1	*TP63*
HOXC4	*BCL2*
PON1	*POLD1*
NRG1	*FGF23*
TP73	*SNCG*
IGFBP3	*GSTP1*
TFAP2A	*STAT3*
SOCS2	*RAD52*
PIK3R1	*TFDP1*
NCOR2	***NCOR2***
ESR1	***TNF***
TSG (11/71)	
BAP1	*ARID1A*
CDH1	*ASXL1*
GATA3	*CEBPA*
PIK3R1	*CREBBP*
WT1	*PRDM1*
	SMAD2
Oncogenes (5/54)	
GATA2	*BCL2*
	DNMT3A
	FGFR3
	U2AF1

B

hypoMRs	hyperMRs
CD34+_vs_AML (6/30)	
(1/8)	**(6/25)**
SEPT9	***DIO3***
	ASCL2
	CDH13
	PTK6
	RUNX3
	SEPT9
Ctr_vs_AML (8/16)	
PITX2	*FSCN2**
ESR1	*GYPC*
CDH1	*KIAA1447**
	RASSF1
	SNX9

C

hypoMRs	hyperMRs
Marcucci *et al.* (22/82)	
CABLES1	*ALOX15B*
CD34	*IQSEC1*
CLDN15	*PRKCZ*
DDIT4	***AATK***
GAL3ST3	***GCNT2***
KLHL3	
MDFI	
ME3	
PRKG2	
WDR16	
APBB1	
ETS1	
F2RL1	
H1F0	
HIVEP3	
KDM2B	
SCRN1	
AATK	
GCNT2	
Li *et al.* (7/16)	
ID4	*TRIM67*
ZFP42	***GPR146***
CHL1	
PCDHAC2	
SOCS2	
GPR146	
Figueroa *et al.* (1/22)	
	LCK

4.5.2.1 Age genes

At least 21 genes (Table 4.5.2A) of the genes in the proximity of these DMRs have been previously correlated to age and are present in the GenAge database[139] (n=307). Some are involved in biologic processes that are hallmarks of aging like DNA damage repair/senescence (*Rad52*, *TP63*, *TP73*) or nutrient sensing/Insulin and IGF-1-signaling pathways (*IGFBP3*, *PIK3R1, SOCS2*). Not present in the GenAge database but also a hallmark of aging were the autophagy genes (for example *ATG16L1*, *ATG5* and *ATG7*).

We further found the expression levels of 3 of these genes could be correlated to survival (Figure 4.5.6), with poor survival attributed to high *SOCS2* expression (p=0.01), high *ESR1* expression (p=5.6x10^{-4}) and to low *TNF* expression (p=0.04).

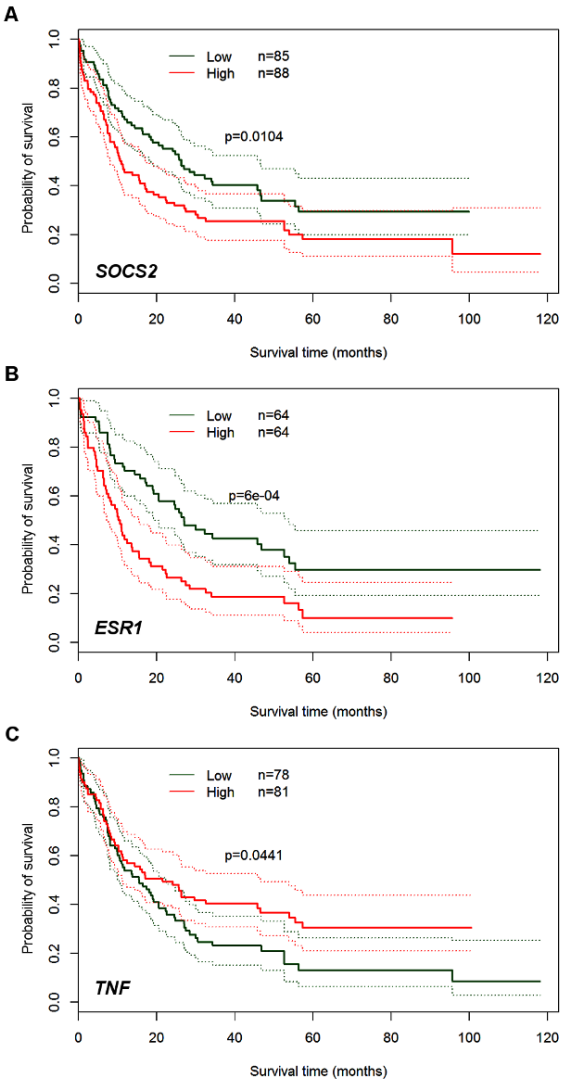


Figure 4.5.6 Overall survival of patient groups defined by high vs low RNA expression levels for age genes that are associated with DMRs.

Kaplan-Meier curves display the estimation of the overall survival for the division of the TCGA cohort into two groups with High (value≥mean) versus Low expression (value<mean) of the genes: *SOCS2* (A), *ESR1* (B) or *TNF* (C), using the expression levels from RNA-seq. The significance of the difference between overall survivals was calculated with the log-rank test. Dotted lines mark the 95% confidence bounds.

4.5.2.2 Cancer genes

Additionally, several genes correlated to hypoMR and hyperMRs of the cluster of the elderly are associated with cancer (Table 4.5.1A). We categorized them using the classification of Vogelstein *et al.*[140] that classified cancer related genes into oncogenes and TSGs using the *"20/20 rule"* whereby *"a driver mutation can be described as oncogenic if at least 20 of its mutations are recurrent as well as being missense"* and *"a tumor suppressor gene would subsequently require a threshold of 20 mutations that all resulted in an inactivating effect."*[140]. Importantly, one known oncogene (*GATA2*) was related to a hypoMR and six TSGs had related hyperMRs, most of them are known for being molecularly altered in AML (*ARID1A*, *ASXL1*, *CEBPA*, *CREBBP*).

4.5.2.3 AML genes

There were also several genes particularly associated to AML related to hyperMRs and hypoMRs of the cluster of elderly (Table 4.5.2B), as determined by matching our list to 2 lists of differentially methylated genes in AML samples when compared to healthy controls. In both studies, the Alvarez *et al.*[88] and the Bullinger *et al.*[137], the number of genes that are related to CpGs exclusively hypermethylated or hypomethylated across all AML samples of corresponding cohorts was very limited (30 of 807 genes and 16 of 80 genes, respectively). Therefore, it was remarkable that 14 of these 43 genes (since 3 genes have both hyper and hypomethylated CpGs) connected to AML pathology were also in the DMRs of the hierarchical cluster of the elderly.

4.5.2.4 Genes related to prognosis predictions in AML

The cluster of the elderly also contained a good number of hypoMRs (n=25, Table 4.5.2C) associated to genes previously reported to have differential DNA-methylation with prognosis correlation (n=120, complete list Table 1.4.1 in section 1.4). Furthermore, we could confirm the poor survival of the patients with high expression levels of *F2RL1* (p=0.04), *HIVEP3* (p=0.02) and *SOCS2* (p=0.01) in the TCGA RNA-seq data, which was consistent with the results in the cohorts of these studies.

While the majority of DMRs found for the cluster of elderly resulted from the higher methylation values found for this group (hyperMRs) not many of these genes were previously mentioned as predictors of survival. Even from the ones that were,

none of them could be confirmed to have prognostic prediction using the TCGA RNA-seq data.

4.6 Robust epigenetic patterns of AML

Since CpG island shores are variable between tissues and very disease-specific we performed unsupervised clustering using the most variable CpGs in island shores (both N-shores and S-shores, Figure 4.6.1). There were several patients with similar methylation profiles that were defined by genetic alterations. There was a first cluster-defined group with 3 samples that have methylation patterns different from all other samples. The next group division (of hierarchical clustering) clearly distinguished *IDH1/2* mutated samples from all others (n=31). After a few more samples were clustered into groups, one of the next group separations divided most of the remaining patients into two groups (dark grey, n=87, and light grey, n=146).

Like in the promoters profile, there was enrichment of elderly patients to the smaller group (dark grey: median age 70y, ranging 41-90y) when compared to the patients in the other group (light grey: median age 57.5y, ranging 21-82y, Welch's t-test Bonferroni corrected $p= 1.54x10^{-9}$). The latter group, with younger patients, contained most of the triple mutated *DNMT3A/NPM1/FLT3* samples (green) and a CBF defined cluster (yellow). Whereas the group of the elderly patients contained samples with complex karyotype/*TP53* (blue) and most of the samples classified as group C using the protein FI network classification.

This group highlighted once more the similarities between the elderly AML samples and samples that are typically known for DNA instability. This resembled what was apparent in the genetic profiles, this time similarities are seen on the epigenetic level.

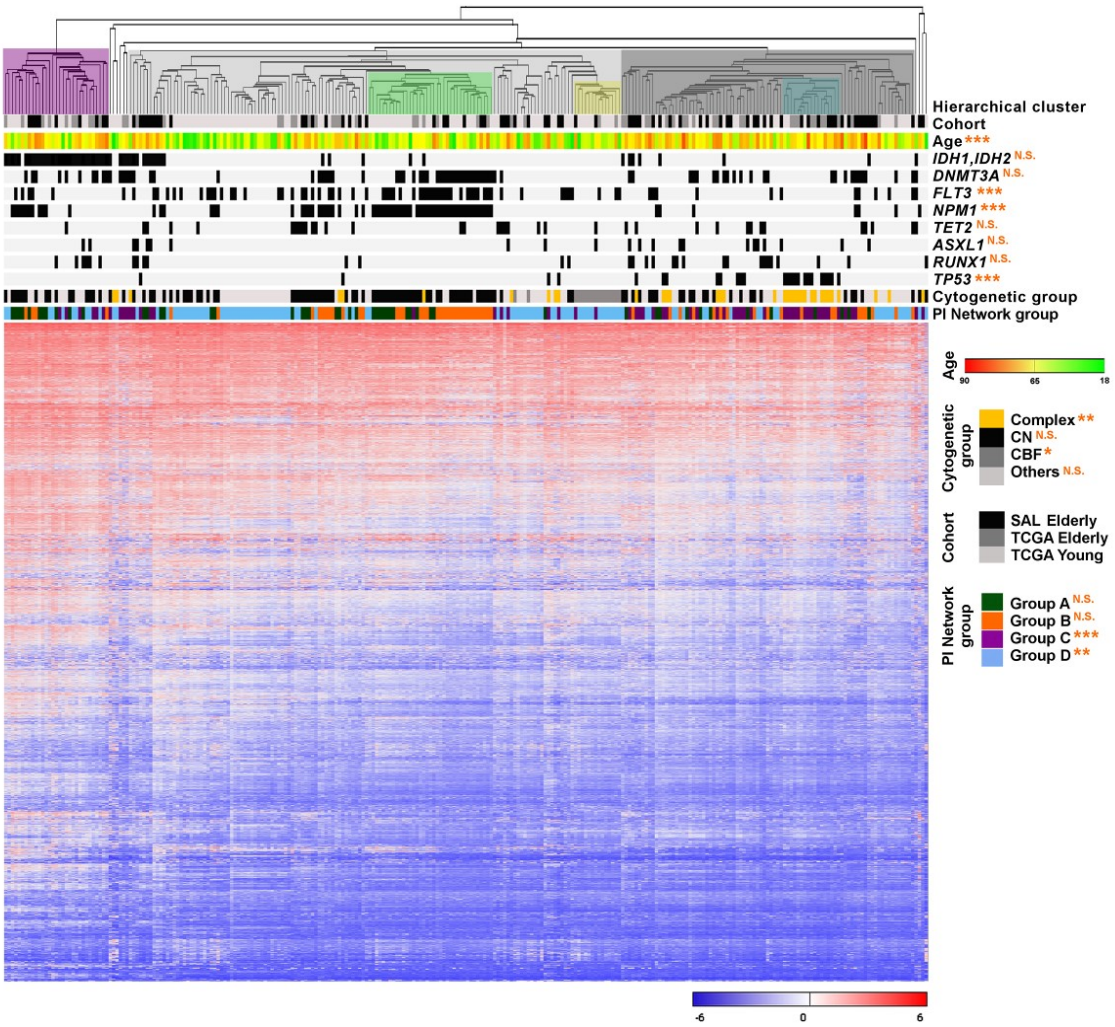


Figure 4.6.1 Methylation profiles of Group C and complex karyotype AML samples and uniqueness of patients with *IDH* mutations.

Analysis of DNA-methylation values in the 6369 most variable CpGs in promoter regions that are in N-shores or S-shores of the CpG islands (in the horizontal axis), scaled from unmethylated (-6, blue) to fully methylated (6, red). Depiction of the unsupervised hierarchical clustering of 274 AML samples (in the vertical axis) with mutation information for high rate mutated genes: 194 patients from the TCGA cohort (136 young of 18-64 years and 58 elderly of 65-88 years) and 80 patients from the SAL cohort (all elderly of 65-90 years). Age of patients at the time of diagnosis was represented as a heatmap code (in the legend). Samples considered part of hierarchical clusters of importance were highlighted. Patients are represented in vertical and classified in cytogenetic groups: cytogenetically normal (CN, no detected cytogenetic abnormalities), core binding factor (CBF, includes t(8;21) or inv(16)/t(16;16)), complex (3 or more cytogenetic abnormalities), or others. *FLT3* included both SNVs and *FLT3*-ITDs available. Adjusted Fisher exact test for skewness of mutations between the cluster in dark grey and the cluster in light grey is specified on the right of each gene, or patient characteristic, or of the legend in case of the groups found using the protein functional interaction network (PI network, N.S. denotes non-significant, *p≤0.05, **p≤0.01 and *** p ≤ 0.001).

4.7 Epigenetic heterogeneity in the SAL elderly AML

In the next step, we dissected the methylome of the SAL elderly AML samples using unsupervised hierarchical clustering (Figure 4.7.1) and identified two discernible clusters (cluster 1 and 2), recapitulating the patterns of the combined cohort. Cluster 1 constituted of *IDH1/2* mutant samples and cluster 2 constituted of triple mutated samples *DNMT3A*/*NPM1/FLT3*. Additionally, a small cluster of samples with *TET2* mutations was observed in this elderly AML cohort (cluster 3).

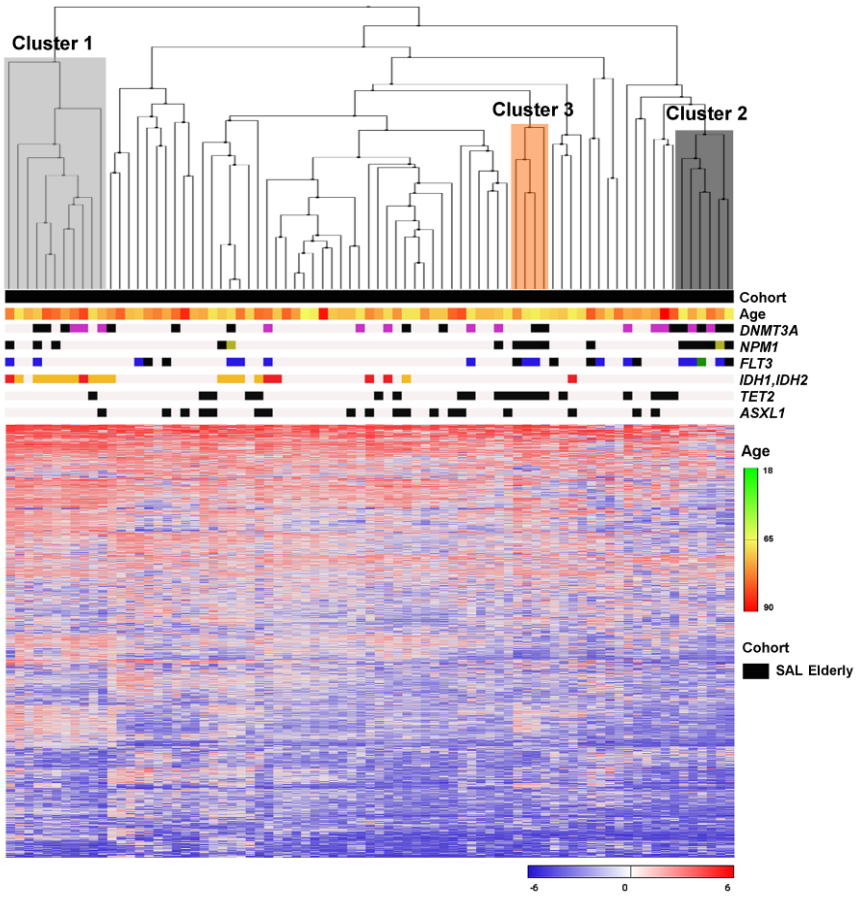


Figure 4.7.1 Methylation profiles of SAL elderly AML patients.
Analysis of DNA methylation values from the 1,000 most variable CpGs of high-dense CpG regions (according to Illumina PHANTOM annotation) using scaled hues from unmethylated (-6, blue) to fully methylated (6, red). Depiction of the unsupervised hierarchical clustering of 79 elderly AML samples integrated with mutation information (for some high rate mutated genes). The different colors represented are: *DNMT3A* R882 mutations (pink), *IDH1* mutations (orange) and *IDH2* mutations (red). Mutational data derived by PCR were included: *NPM1c* mutation status (light green), *FLT3*-ITD (blue) and concomitant *FLT3* SNV with *FLT3*-ITD (green). Samples considered part of specific clusters were shaded and clusters labeled 1 to 3. Age of patients at the time of diagnosis was depicted as a heatmap code (in the legend). Adapted from Silva *et al.* 2017[1].

We obtained DMRs in two-group comparisons (cluster vs non-cluster samples and mutated vs wild-type samples). This analysis showed definite patterns of differential methylation (Figure 4.7.2A). Cluster 1 (and *IDH1/2*) showed about 230

DMRs, almost all hypermethylated, associated with genes enriched for biosynthetic/metabolic processes and myeloid cell differentiation (Figure 4.7.2B). For cluster 2 (*DNMT3A*/*NPM1*/*FLT3*) and for *DNMT3A* we found regions were exclusively hypomethylated, particularly affecting Homeobox genes (*HOXB3, HOXB4, HOXB5, MEIS1,* Figure 4.7.2).

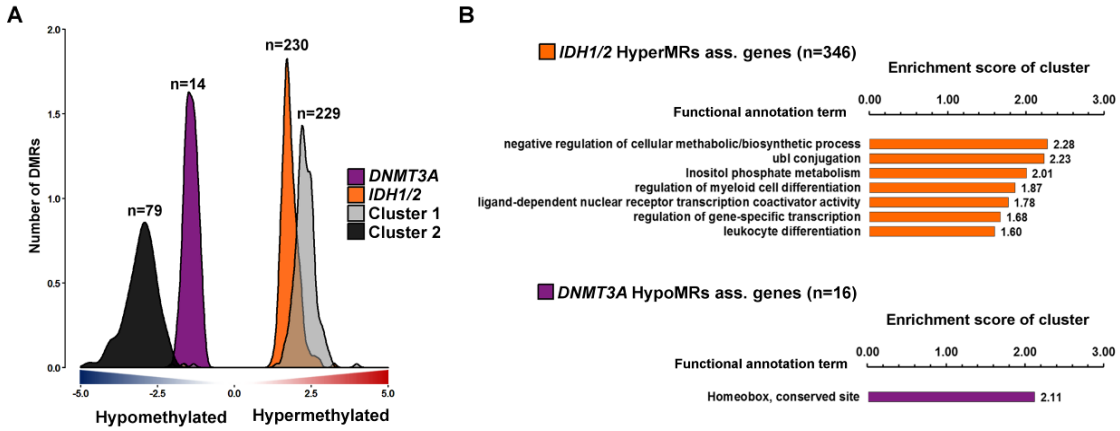
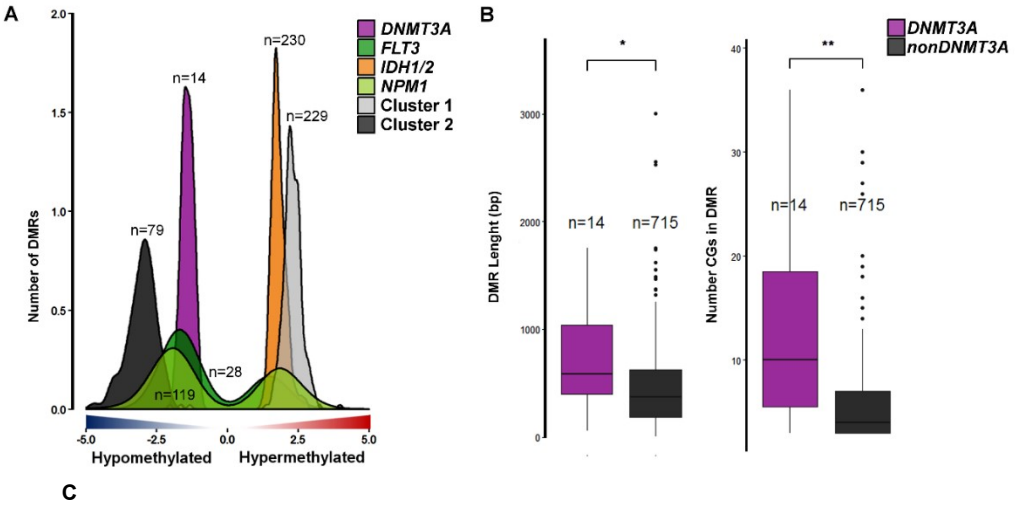


Figure 4.7.2 Specific differential methylation signature of epigenetic regulators mutated in the SAL elderly AML.
(A) The density of differential methylated regions (DMRs) plotted for mutational status groups of mutant *IDH1/2* or *DNMT3A* (versus its wildtype samples) and for methylation cluster 1 or 2 from the unsupervised analysis (against non-cluster samples). The numbers of DMRs with at least 3 CpGs differentially methylated were obtained from the most variable 104,852 CpGs and classified into hypermethylated regions (hyperMRs) or hypomethylated regions (hypoMRs), according to their ratio of methylation in the test group (n is the total number of DMRs). (B) Biological terms found enriched in the list of genes associated with *IDH1/2* hyperMRs and *DNMT3A* hypoMRs are enumerated (n ass. genes was the number of associated genes). Associations were obtained using GREAT[125] and enrichments score for each functional annotation cluster were calculated using DAVID (EASE scores with FDR<10%)[126,127]. As presented in Silva *et al.* 2017[1].

However, *NPM1* mutated or *FLT3* mutated samples by themselves did not share this clear tendency, having only slightly more regions classified as hypomethylated than classified as hypermethylated (Figure 4.7.3A). Like in traditional cancer DMRs, for both of these the hypermethylations were more correlated to gene promoters than the hypomethylations (Figure 4.7.3C).

In comparison, *DNMT3A* mutations despite being present in many samples with *IDH1/2* mutations do not show any hypermethylated regions. Furthermore, hypomethylated regions in *DNMT3A* mutated samples were distinctly covering extended regions of the genome, being bigger than the average other DMRs, mostly due to larger numbers of CpGs changed by region (with the average of 10 CpGs changed in each region, Figure 4.7.3B).



Characteristic in test group	Cluster 2	*DNMT3A*	*NPM1*	*FLT3*	*IDH1/2*	Cluster 1
Samples (of 79)	6	28	18	22	21	11
DMRs	79	14	119	28	230	229
HypoMRs	79	14	72	20	1	3
Genes associated to HypoMRs	101	23	83	26	2	6
HypoMRs in a gene[1]	66 (84%)	13 (93%)	58 (81%)	19 (95%)	1 (100%)	3 (100%)
HypoMRs in a promoter[2]	17 (22%)	5 (36%)	18 (25%)	3 (15%)	1 (100%)	1 (33%)
HypoMRs in a TSS[3]	9 (11%)	2 (14%)	9 (13%)	1 (5%)	1 (100%)	0 (0%)
HyperMRs	0	0	47	8	229	226
Genes associated to HyperMRs	0	0	81	26	352	337
HyperMRs in a gene[1]	0 (0%)	0 (0%)	43 (91%)	8 (100%)	187 (82%)	186 (82%)
HyperMRs in a promoter[2]	0 (0%)	0 (0%)	22 (47%)	7 (88%)	109 (48%)	80 (35%)
HyperMRs in a TSS[3]	0 (0%)	0 (0%)	10 (21%)	2 (25%)	40 (17%)	38 (17%)

Figure 4.7.3 Specific signature of differential methylation for mutated genes in the SAL elderly AML.

(A) The density of the significant differential methylated regions (DMRs) for groups of mutant versus its wildtype samples (for *IDH1/2*, *NPM1*, *FLT3* and *DNMT3A* mutations) and for the methylation clusters against non-cluster samples. DMRs were classified into hypermethylated DMRs (hyperMRs) or hypomethylated DMRs (hypoMRs) according to this methylation ratio. (B) Length of the DMRs and number of CGs they contained, for the DMRs of *DNMT3A* mutants vs all the DMRs found for all other groups tested (*nonDNMT3A*: *IDH1/2*, *NPM1, FLT3, TET2, ASXL1, RUNX1*, *SRSF2*, and clusters 1,2,3). n: the total number of DMRs for each comparison group; *p≤0.05; **p≤0.01. (C) The table reports differently methylated regions in specific groups of the SAL elderly AML. DMRs relate the number of highly significant DMRs (with at least 3 CGs changed) for high rate mutated genes or clusters from the unsupervised analysis. The DMRs and the genes found to be associated with these regions were classified according to each state of differential methylation (hypoMRs or hyperMRs). The number of DMRs associated to specific regulatory features are stated with the percentage they represent: 1- DMRs with at least 1b overlapping a gene; 2- DMRs within 1kb of a gene promoter; 3- DMRs with at least one 1b overlapping to a transcription start site (TSS). Associations were calculated by EpiExplorer[128].

CHAPTER 5. DISCUSSION

Since the advent of NGS, we have recognized that AML has a large heterogeneity in molecular alterations that make it a highly complex disease, but its patterns are resolvable for the number of mutations found in AML (average 13 Tier1 mutations) is not as high as in most of the other cancers[141]. The recent NGS studies suggest the development of an AML involves the acquisition of somatic mutations in a stepwise manner, due to the accumulation of mutations in stem cell progenitors responsible for continuous blood production. This dynamic of the disease would explain why AML is more frequently found in the elderly population.

Among the molecular alterations found in AML many occur in epigenetic regulators, with a high prevalence of mutations found for example in *IDH1*, *IDH2*, *DNMT3A* and *TET2*, which underscored possible roles of the epigenome in the pathogenesis of AML. The initial observations that epigenetic mechanisms are dysregulated in AML lead to the analysis of the unclear relationships between patterns of mutations and epigenetic phenotypes[37,79,66]. Nowadays these patterns can be comprehensively explored since both can be obtained with whole genome/exome coverage.

At the time of our study the molecular analysis of AML, genomic and epigenomic landscapes, referred either to selected groups of patients, like cytogenetically normal (CN[66]), *de novo* or secondary AML (sAML[49]), young patients (<60 years old[79]), or to unselected cohorts like the one with 200 samples sequenced by TCGA[37]. Despite the exponential increase of studies in genetic or epigenetic classifications of AML patients in the last 2 years, both patterns had not been addressed together specifically in the elderly patients (using either global or non-global approaches), until our study[1].

Therefore, we aimed to unravel the genetic and epigenetic patterns of the elderly AML that could provide more detailed evidence into its specific pathogenesis. We described many specificities found in the SAL elderly cohort and our procedures allowed us to point to known and new molecular alterations of biological relevance or prognostic value that can be used as druggable targets.

5.1 Genetic patterns of elderly AML

In our comprehensive approach, we analysed a large set of candidate cancer genes (555) by targeted NGS and established several particularities of the elderly AML genetic profiles.

With this extended panel sequencing, we found 281 genes mutated, which was about 5x more genes than included in the panel Illumina has set up as the myeloid sequencing panel (54 genes). Since the elderly patients of TCGA (≥65 years old) showed that the median mutations per patient was high (when compared to the younger patients) panels like this would have proved insufficient to evaluate AML in the elderly population.

5.1.1 Molecular alterations correlated to age

The number of mutations observed per patient did not directly correlate with the age neither in the SAL elderly nor in the TCGA elderly, even though the increased age related to decreased survival probability. Instead, the increase in the median number of mutations per patient could only be seen between TCGA young and TCGA elderly group of patients. Both facts were consistent with young and elderly AML being distinct groups of AML.

This also indicated that specific genetic alterations and patterns in elderly AML would have to be analysed for only they might provide novel insights for the dismal prognosis of these patients.

With our approach, we obtained **extensive information** that was not captured as a whole in other studies investigating elderly AML. While others describe increased frequencies of certain mutations in older patients[131,142,143], we were able to extend this mutational pattern by exploring phenotypic interactions between functional groups of proteins. The most prominent characteristics of the genetic patterns of elderly AML to emerge were: (i) that almost all elderly patients had at least one mutation in an epigenetic regulator (85%), (ii) the large percentage of patients with mutations in splicing regulators (38%) and (iii) the presence of mutations in several proteins involved in DNA repair.

The high coverage of targets obtained in the many **epigenetic regulators** we targeted could be partially responsible for the high amounts of alterations detected in the SAL elderly cohort. Since these many of targets had only been found by the TCGA study, and because it was done using a whole-exome approach they have less coverage, there were difficulties in effecting comparisons. Hence we looked for further confirmation, which was then obtained by analysis of the young vs elderly patients in the TCGA cohort. Furthermore, the increased frequency of mutations in epigenetic factors was not completely unforeseen, since many of these mutations had previously been correlated with increasing age of AML patients[28, 131,142,144].

Still, the amount of these mutations that could be identified was prominent for together they are present in the majority of patients. One example of this was the unexpectedly **high amount of *IDH1* mutations** (17%), a fact for which age could be blamed since no other explanation could be retrieved. To our knowledge this has not been confirmed in other cohorts, usually, *IDH2* is the enzyme with more mutations (between the two) and the one that has been correlated to older AML patients[145,146,147]. Another example was the **high frequency of mutations in *ASXL1*** (22%) that was previously found at 6% in a young AML cohort[132] and at 11% in *de novo* AML[148]. The latter study correlated *ASXL1* mutations to age for their occurrence in 6% of young patients (<60y) and in 18% of older patients (≥60y)[148]. Similarly, in a cohort with CN-AML patients, *ASXL1* mutations were present in 3% of the young patients (≤60y) and 16% in the older patients (>60y)[142]. A further example was the 26% of the SAL elderly AML patients with a *TET2* mutation. This is considered a **high frequency of *TET2* mutations** because these have been reported in age unselected cohorts of *de novo* AML at 8%[37] and 13%[131]. This second study also concluded these were closely associated with older age as they were present in 7% of the young patients (≤60y) and 24% in the older patients (>60y)[131]. Furthermore, new studies that applied the most recent targeted NGS analysis (which we used for comparison in results section 4.2) have also confirmed the substantially increased incidence of these mutations with age[34,38].

It was striking to find a high frequency of mutations in **spliceosome components**. We found unusual the number of mutations in *SRSF2* (23%) since at the time there was no comparable value. After our first report[2] other studies have found that the frequency of mutations in *SRSF2* reported by the early studies (~1%) was much lower than the ones found by panel NGS nowadays (6% or 10%)[38,39]. Additionally, Eisfeld and colleagues[34] have reported a similar value (~21%) when looking at their group of older patients (≥60y). We believed this was explained by the low coverage of previous studies, which is believed to be due to the high CG content of the region with the most frequent mutation (Proline 95), this was also the assumption made by Papaemmanuil and colleagues[39] (when comparing to TCGA).

Furthermore, using our extensive target list, we identified several mutations in genes related to **DNA repair** that were not restricted to *TP53*. These genes included other DNA damage response and surveillance proteins such as BML, BRIP1, XPC, ERCC2, ERCC3, ERCC4, ERCC5, BRCA2, FANCA, FANCC, FANCD2OS, FANCF, ATM, CHEK2, NBN, MSH6 and MLH1 (many of these mutations have been found in other tumor types). Furthermore, *TP53* was frequently mutated in SAL elderly AML (10%), a reasonable

expectation since an age association for this gene could be found in the TCGA cohort (increasing from 4% in TCGA young to 18% in TCGA elderly).

5.1.2 Factors for elderly AML poor prognosis

In the elderly AML, there are typically high frequencies of cytogenetic patterns of poor prognosis, which might help explain the dismal outcomes of these patients. The SAL elderly AML had rates of **CN-AML** not very different from younger cohorts. In turn, **CBF** samples or balanced abnormalities (of favorable prognosis) were not as frequent as in young, but high frequencies of **complex karyotypes** and **unbalanced rearrangements** (of unfavorable prognosis) were seen. Several papers[34,40] published a bit before and right after our report[1] confirmed the congruence of this SAL elderly AML cytogenetic distribution. This includes the comparable amount of CN, the increase in complex karyotype and the low amount of CBF found in older patients that were later corroborated by Eisfeld and co-workers[34].

Since **cytogenetic characteristics** have been insufficient to explain elderly AML poor prognosis[106,107] we had postulated that **specific genetic alterations** could correlate to the poor prognosis of these patients.

We found the main factor of elderly AML poor prognostic could be related to the frequency of mutations in specific genes, since we identified a high frequency of mutations in genes known as **poor prognostic predictors**: *DNMT3A*, *SRSF2*, *IDH1/2*, *RUNX1*, *TET2*, *ASXL1*, *TP53*, *BCOR* and noted the low frequency of mutations in genes typically correlated with **favorable outcomes**, *NPM1* and *CEBPA*.

We described this by comparing SAL elderly values to two particular studies, Metzeler and colleagues[38] and Eisfeld and colleagues[34]. Although both sequenced using different NGS panels with smaller sets of genes, limiting some comparisons, they included many samples. We compared our resulting frequencies to both these studies keeping in mind each one has its own peculiarities in relation to ours. In the study of Metzeler and colleagues[38] only 68 genes were sequenced but 31 of them only in known hotspots. In the study by Eisfeld and colleagues[34], the samples were restricted to *de novo* AML and only 79 genes were sequenced using an NGS procedure.

There were several other **technical considerations** that advised for caution in these comparisons. Many of the genes sequenced in ours and these 2 studies do not have high mutation frequencies and do not allow proper analysis of the differential amount of mutations. Hence, there were low frequencies (which were in agreement with one or both this studies) that we included in results but we did not stress for fear these might be resulting of unknown selection bias due to the small number of patients in the SAL elderly AML, like *KRAS*, *NRAS, ZRSR2*, *GATA2, RAD21,* and *WT1*. Furthermore, some

mutations were not completely ascertained in SAL elderly AML and were not used, like *FTL3*-ITD. These eluded our attempts of determination using the Pindel algorithm and manual curations were not clear. We were confined to use the data provided by the diagnostic testing, obtained for 70 of the 93 patients. Similar problems do not allow the comparisons of the frequency of mutations in *MLL* since *MLL*-PTD is the most frequent mutation. Some new algorithms and changes to Pindel have been developed meanwhile[39,149,150] and they might make it possible to include large mutations like these in a future analysis.

We mostly found tendencies of increasing or decreasing frequency of mutations with patients' age that have now been observed in other cohorts, except for the mutations in *NPM1*. There was an appreciable **low amount of *NPM1* mutations** found in the SAL elderly (16%) by using our strict criteria of considering only mutations in areas where the coverage was 30x and VAFs higher than 20% obtained by NGS. Other studies have found *NPM1* mutations much more frequent, age unselected cohorts having about 30% to 34% of mutations in *NPM1*[38,34]. Since the tetranucleotide insertions (known as *NPM1c*) are prone to be missed due to the low coverage on this small insertion position, we did a manual evaluation of all samples and retrieved 2 mutations that were missed due to the low amount of fragments mapped to the region. The evaluation of *NPM1c* amplified by PCR and Sanger sequenced reclaimed another 2 mutations, that were not even visible in NGS, bringing our maximum number to 20 mutations (20/93=22%). Therefore, the low number of *NPM1c* mutations remains unexplained and a correlation of this gene with age is not established by others. Then again, it seems plausible this frequency would be low in elderly AML because these mutations do not usually co-occur with mutations in *ASXL1* and *RUNX1*, which are both increased in the SAL elderly AML.

The survival prognosis for patients with mutations in single genes (or combinations) that are **well-established survival factors** in non-elderly specific cohorts did not provide survival predictions in SAL elderly AML, except for the two strongest poor prognostic factors **complex cytogenetic** and the ***TP53* mutated**. On the one hand, our low power for these predictions might be a reason for this fact. On the other hand, other studies have also observed the **loss of several predictions in the older cohorts**, when using factors that have been seen in age unselected cohorts and in young cohorts. In their study, Tsai[40] and collaborators lacked the survival prediction of the double-mutation in *CEBPA*, the *RUNX1* and the *IDH2* mutations when using only the older patients. Their elderly retained only the good prognosis of *NPM1* mutated/*FLT3*-ITD wild-type vs other subtypes and the bad prognosis of *DNMT3A* and of *TP53* mutated. In contrast, in the study of Metzeler *et al.*[38] the bad prognosis of *DNMT3A* mutations was only observed in the young group and

the good prognosis of *NPM1* mutations was only seen in the older group. As a result, the combinations of mutations in *NPM1*/DNMT3A only had predictive value in the group of young patients. Additionally, the poor survival prediction of *RUNX1* mutations was also not observable in their group of older patients, while the good prediction of *biCEBPA* and the bad prediction of *TP53* mutated were observed independently of the age group[38]. Therefore, it was not unthinkable that only the stronger prognostic factor seen in both these studies (*TP53* mutated) would show significant impact in survival probabilities in the SAL cohort.

A prognostic factor not lost in SAL elderly AML was the **WBC count**, which was a poor prognosis predictor at values as low as 35/nL, even when hyperleukocytosis is only deliberated for values superior to 100/nL. When using the WBC levels to divide the SAL elderly into two groups with different survival probability it revealed a correlation of WBC≥35/nL to the presence of *NPM1* or/and *FLT3* mutations in samples and WBC<35/nL to the presence of *IDH1* mutations. This exposed the diverse nature of the two groups, possibly reflecting a higher proliferation rate of the disease with *NPM1* or/and *FLT3* mutations than with the *IDH1* mutated. A difference that would be reflected on the low survival probability of the high WBC group (WBC≥35/nL). This opposition between samples with mutations in *IDH1* vs *NPM1*/*FLT3* highlighted the heterogeneities that could be found amongst the SAL elderly AML.

5.1.3 Genomic classifications of elderly patients

We had indications elderly AML had **specific genetic patterns** due to the increased frequency of mutation in elderly AML, but the heterogeneity of the SAL elderly AML was not entirely different from younger cohorts. Elderly AML patients had known patterns of co-occurrence and mutual exclusions of specific mutations. The mutations in epigenetic regulators (both *IDHs* vs *TET2*) still mainly appeared once per patient and co-occurrence of mutations in splicing components was also nearly null. Elderly AML could also be divided into groups of **patients bearing similar genetic profiles** and these groups were quite relatable to the ones found in younger cohorts.

Using the new genetic-based classification of AML, proposed by Papaemmanuil and co-workers[39], we observed that the majority of our elderly AML patients fell into their class of **"AML with mutated chromatin/splicing regulators"** (instead of to class "*NPM1* mutated"). There were 34.4% of our patients that belonged to this class, with co-occurring mutations in *SRSF2*, *SF3B1*, *U2AF1*, *ZRSR2*, *ASXL1*, *BCOR*, *MLL*, *EZH2*, *PHF6* and *RUNX1* (p=0.006). Consistent with our results, in their study[39] this class (only 18% of their cohort) was correlated with older age but not with sAML samples. The prominence of these mutations was news at the time, but have now been confirmed in the mutational oncoprints

of several elderly cohorts[34,38,151]. For a time most of these mutations were referred to as "secondary AML mutations" but according to our analysis (and in those studies also), the clinical definition of sAML does not reproduce the biological entity where these are present.

As previously mentioned, ***TP53*/aneuploidies** were also found in high frequency in elderly AML and this was the other genetically defined group that had been correlated to age[39]. These particular mutational patterns are said to develop due to DNA instability of the stem cell progenitors since they are related to impairments of DNA repair. This characteristic of this AML class is pointed as the reason these clones are particularly difficult to eradicate.

In opposition, the ***biCEBPA* group** was not detected in SAL elderly AML and was 4% in the age unselected cohort used by Papaemmanuil and co-workers[39] to establish the genomic classification. In the SAL elderly cohort, *CEBPA* mutations were present but there was not homozygous and were present in different patients. This could be coincidental because the number of *biCEBPA* was expected to be low, any accidental bias in the SAL cohort could be responsible.

If the **general increase of mutations in elderly AML** was a product of random occurrences the many patterns of genetic classifications that are emerging in the younger cohorts would not have been present in elderly AML, which was not the case. This analysis pointed to the redundancy of mutations within the similar molecular functions and the possible molecular synergy among specific mutations, which was worth exploring.

5.1.4 Functional interactions of protein targets

Having identified 281 mutated genes in elderly AML we used clustering algorithms to group the genes into modules based on protein-protein interactions. This methodology enabled us to unravel pathway information using the functional relation between the proteins affected by mutations.

This exploration gave new insights, as it separated proteins into **groups with different functional properties**. These sub-networks identified molecular signatures of patients, with different overall survival probabilities, based on their mutational patterns in 2 of these sub-networks: NPM1/mRNA processing proteins or DNMT3A/DNA repair proteins.

The close relation of **DNMT3A and DNA repair proteins** that brought them into the same functional interaction module suggests a possible interaction of DNMT3A in DNA repair mechanisms. This possible functional role of DNMT3A in DNA repair has been recently explored by Guryanova and co-workers[152], who showed impairment of DNA damage sensing mechanisms in *DNMT3A* mutated cells was a possible explanation for the treatment resistance observed in AML.

In contrast, the patients with ***NPM1* mutation and/or splicing regulators** seem to be less resistant to treatment in SAL elderly AML. A fact that is in line with the reports of favorable treatment response of elderly patients with *NPM1* mutations in groups of patients more than 60 or 70 years old (higher complete remission rates and longer overall survivals)[38,103], as we have mentioned. This was the only favorable prognostic factor found to be stronger in the elderly than in younger patients, has been validated in several other cohorts and found to be of special importance in the elderly AML[38,153,154]. We speculated that because we had a good detection of *SRSF2* mutations (and a high number in elderly AML) the patterns of these groups became clear in the SAL elderly AML, despite our low statistical power.

We expected this biological characteristic to be **robust** and the result **reproducible**. In the TCGA cohort, we could replicate the main result by determining the bad prognosis of samples with *DNMT3A*/DNA repair mutations lacking mutations *NPM1*/mRNA processing proteins. This was attainable despite their low number of *SRSF2* (0.5%) and regardless of the composition of the cohort being restricted to *de novo* AML. Furthermore, the disparity of the survival probability of patients with mutations in DNA repair and *NPM1*/mRNA processing can also be evaluated by observing the decreasing survival probability of the 3 different genetic classes: "*NPM1* mutated" vs "chromatin/splicing regulators mutated" vs "*TP53*/aneuploidy" (see Figure 1.3.3) in the cohort from Papaemmanuil and co-workers[39].

5.1.5 Mutational progression into elderly AML

Last, we took notice that elderly AML mutation rates appeared to **reflect pre-existent alterations**, bearing high mutation rates in the chromatin, epigenetic and splicing regulators most of them **found in CHIP**. Since the genes, *DNMT3A*, *TET2*, *SRSF2, ASXL1*, *SF3B1, IDH1/2,* and *TP53* had increased frequencies of mutation in elderly AML and have previously been related to clonal hematopoiesis in the healthy elderly population[10,11]. Moreover, these somatic mutations (in *IDH1*, *IDH2*, *TP53*, *DNMT3A*, *TET2* and spliceosome genes) were recently confirmed to be part of the **premalignant mutational landscape**, being present years before the development of AML and significantly increasing the risk of developing the disease[155]. Also of note, not all genes formerly detected to be mutated in CHIP presented with an increased mutation rate in elderly AML (for example *GNAS* and *JAK2*). The **lack of enrichment of mutations in *JAK2*** in the elderly AML was especially surprising since this is a frequent mutation in CHIP (as frequent as *IDH2*, *TP53* or *ASXL1*, depending on the study). Although, in these studies, the mutations in this particular gene failed to show a trend to increase with age, while *TET2*, *ASXL1* and *SF3B1* were predominantly found in the oldest individuals with CHIP[11].

The presence of mutated epigenetic regulators in CHIP and elderly AML indicates the epigenetic changes might be preceding AML driver mutations in proliferative genes and contributing to the development of hematopoietic diseases.

Finally, in the elderly AML, some mutations in *DNMT3A*, *TET2, TP53,* and *RUNX1* had very high VAFs which denoted that in many blasts these were homozygous mutations (marked as LOH). This could be an indication that these mutations had **occurred very early** in the elderly AML development. In this case, it is important to highlight that *RUNX1* mutations are typically found in MDS but not usually found in CHIP[11].

All of this leaves unanswered questions about the course of development of elderly AML, for it can either evolve from undiagnosed MDS or develop directly from CHIP. But it definitely pointed to a correlation of the elderly AML disease to the increased epigenetic variability of HSC with age.

5.2 Epigenetic pattern of elderly AML

Since in general the methylomes are impacted by age[156] it was important to address the specificity of the elderly AML methylome, for this changes could contribute to the heterogeneity of the disease.

Thus, we compared the epigenetic profiles of elderly AML with a group of younger patients. We combined the methylation profiles of our cohort with the TCGA cohort methylation profiles, providing power for the comparisons between the two age groups. Due to the robustness of the Illumina 450K assay and the available normalization algorithms we experienced **no difficulties in integrating the data** we obtained for SAL elderly AML with the TCGA data.

We segregated patients into a group of elderly AML (≥65years) and a group of young AML (<65years) and used these to determine DMRs. Elderly AML revealed a low number of DMRs showing no strong bias for hypermethylation vs hypomethylation, exposing the **moderate effects of the categorical age** in the AML epigenetic pattern. The elderly vs young AML had several DMRs located -5kb to 1kb distance from gene promoters. Elderly patients had lower levels of DNA methylation in 244 genes, from which 12 were related to cellular carbohydrate metabolic processes (GO:0044262), and had higher levels of DNA methylation in 91 genes, from which 17 were functionally related to biological adhesion (GO:0022610). Furthermore, both these DMRs have many genes annotated to regulation of metabolic process (GO:0019222).

Although these biological functions have been related to aging[24] in our analysis of the DMRs of the elderly AML **we did not find the age-related CpGs** referenced in other reports[157,158,159]. These age-related CpGs were CpGs with clear correlations between DNA methylation levels and donors age that others found using data from whole blood samples[157,158,159]. In fact, it was shown that age-related CpGs suffer an almost linear hyper- or hypomethylation that allowed the construction of models using them to estimate chronological age[159]. We particularly did not find the three CpGs from the model in Weidner *et al.*[160] that demonstrated these were enough to estimate biological age in blood.

Upon further analysis, we found that several of our unsupervised hierarchical clustering of these epigenetic profiles resulted in the separation of patients into two groups with strong statistical differences in age. This showed that the elderly AML patients have **distinctive epigenetic patterns associated with age**, which was a strong feature in the analysis of the CpGc in promoters. The group of patients with the majority of elderly samples, formed in this manner, was denominated cluster of elderly and used to probe the differences between these AML diseases. The analyses showed that epigenetic patterns of AML **decoupled from age according to patterns** related to genetic characteristics.

We observed the specific age-related signature of DNA methylation at promoters was significantly correlated to several mutations, being positively correlated with mutations *IDH1/2*, *RUNX1* and *ASXL1*, as samples with these mutations were almost exclusively in the cluster of the elderly.

These previous observations were in line with the report of Lin and Wagner[157]. They did a systematic examination of age-associated DNA methylation patterns of 25 cancers using TCGA data[157]. The results clearly indicated that epigenetic patterns of the **categorical age were abrogated in cancers tissues**. They found AML, in particular, lacked any CpG methylation levels with a good correlation to patients' age[157]. They further report that CpGs hypermethylated upon aging in non-malignant tissues were coherently modified in a non-stochastic manner in AML depending on mutations[157]. Higher **age-predictions were associated with mutations** in *RUNX1*, *WT1*, and *IDH2*, and younger age-predictions were predicted in *TET2*, *TP53*, mutations and *PML-RARA* fusions[157].

Knowing this, we used the cluster of the elderly as the **epigenetically defined group of elderly AML** for further explorations. We noticed the cluster of the elderly AML had more hyperMRs than hypoMRs, but in general, in the whole blood of older age healthy donors, the number of hypomethylated CpGs supplants hypermethylated CpGs[157]. Furthermore, the hyperMRs of the cluster of elderly had no clear relation to promoters but the hypoMRs were enriched in promoter regions (with 61% of the hypoMRs while in the 450K assay only 20% of the CpGs are annotated to promoters). While for whole blood age-related hypermethylations were the ones significantly enriched in gene promoter regions, whereas hypomethylations occurred rather outside of gene promoters[157].

5.2.1 Pathways involved in elderly AML epigenetics

The combined cohort revealed a specific elderly AML epigenetic signature (quite dissimilar from young AML) that was not just driven by physiologic changes in age-associated CpGs. In turn, genes potentially affected by differential methylation between the elderly and the young were enriched in **cellular components connected to age** (for example extracellular matrix and cytoplasmic vesicles components) and in **genes connected to AML phenotypes** (mainly *HOX* genes and myeloid differentiation regulators).

This distinction of the DNA methylation changes that are attributed to age or attributed to cancer is not clear because of the **overlap between age-associated** CpGs and **cancer-associated** CpGs[161]. After an effort to dissect this effects Wang *et al.*[161] concluded: *"Genes only differentially methylated in aging[(i)] were mainly enriched in Golgi vesicle transport and regulation of cellular metabolic process. Genes only differentially methylated in cancer[(ii)] were enriched in immune response, defense response and*

response to wounding. While overlapped[(iii)] *genes were primarily involved in cell-cell signaling, cell adhesion and neuron differentiation, etc."* [161]. Our analysis found many gene enrichments with biological/cellular functions related to the ones in their report: (i) regulation of biosynthetic/metabolic process and cytoplasmic vesicle (aging); (ii) inflammatory response, phagocytosis and endocytosis (cancer); (iii) cell-cell signaling, synapse and cell junctions, forebrain and hindbrain development (overlapped).

The genes related to the DMRs in our list could be responsible for elderly AML specific phenotypes **if their expression is altered in the elderly** vs the young patients.

However, none of them were reported in the work of Lin and Wagner[157], which had a very **short list of genes** (n=11) **differently expressed** between two age groups (elderly when compared to the young), obtained by using the RNA-seq from the original TCGA cohort. Ten of them were higher expressed in the older group (*C7orf13*, *CLU*, *DSC2*, *FAM127A*, *FAM127B*, *JAG1*, *LOC644538*, *NHLRC1*, *TEKT2*, and *TTC12*) and only *NEXN* (Nexilin F-Actin Binding Protein) was less expressed. Interestingly from those 10, only the gene *C7orf13*[*MY040*] was present, but in our list of genes with hyperMRs in the elderly. This RNA gene from the class of non-coding RNAs has recognized involvement in the regulation of endocytosis[136]. The phenotype of low expression of *C7orf13* (achieved by siRNA in HeLa cells) is decreased transferrin endocytosis and decreased endosome-nucleus distance (endosomes clustered in the perinuclear region)[162]. We observed that for many genes with marked high methylation levels in the elderly AML this was a phenotype attributed to their low expression.

In addition, another study dissected the **age-specific biology of AML** by analysing the gene expression of gene-sets that constitute signatures of specific pathways[108]. Applying this methodology, Rao *et al.*[108] determined older patients with AML had a **lower probability** of E2F and PI3K pathway activation but a **higher probability** of RAS, TNF, Src, and epigenetic stem-cell signature pathway activation[108]. These were not the pathways found to be especially enriched in our lists of genes with DMRs for the elderly AML (compared to younger AML). Contrary to the Rao *et al.*[108] study, our result found *TNF* to be hypermethylated in the elderly AML samples, which indicates the probability was for low activation of the TNF pathway in this samples.

These **lack of direct correlations** were an indication the relation between gene expression and DNA methylation is not easily deducted in AML, a fact that is a well-known problem, usually attributed to the high complexity of gene regulation processes.

5.2.2 Regions, genes and prognosis predictions of elderly AML

We thought the distribution of DMRs across chromosomes could hold some information about the epigenetic difference between elderly and younger samples. We

found most regions with very high methylation levels in samples of elderly patients (hyperMRs) were **localized at the ends of chromosomes**. This result was thought-provoking since we could not find a good explanation for the observation, two hypothesis could be put forward.

The first idea was, the tips of chromosomes in the elderly AML could be protected by **shorter telomeres** and therefore be different in elderly patients when compared to young. This could be due to telomere shortening in the aged progenitor cells[163] that give rise to the elderly AML. The telomeres have been found to be involved in loops with genes located up to 10Mb from the tips of chromosomes of human myoblasts and fibroblasts[164]. This 3D conformation of DNA is modified when the telomere changes in length, affecting the expression of genes in the region[164]. Since the position of the hyperMRs in chromosome tips was mostly within these 10Mb regions this could indicate we were picking up on different DNA conformations as a result of telomeres shortening.

Another possibility was, that the elderly AML blasts have compromised **boundaries of subtelomeric regions**, which again could be coming from aged progenitor cells that give rise to the elderly AML. A fact with no precedent, as the existence of a boundary to the subtelomeric regions was only recently postulated in *S. pombe*[164], where it was found to protect the genes in the tip of chromosomes from a heterochromatin spread, which results in gene silencing[164]. Since our hyperMRs regions were not restricted to subtelomeric regions (telomere-adjacent regions of approximately 500Kb[165]), but they seem to be mostly concentrated in the immediate proximity of subtelomeric regions, this would be a plausible reason.

Taking a further look at the hyperMRs in the elderly AML group there was a region within chromosome 17 that called to attention. Not only for its **uncommonly big size** but also because it overlapped several genes associated to **cellular processes of metabolism, endocytosis and cell adhesion**, many of these proteins being associated to the organization of the actin cytoskeleton. This was remarkable for these were the 3 processes that were strongly implicated in **stroma–leukemia interaction**[26]. They were found to be deregulated in both the transcriptional and the DNA methylation signatures of the mesenchymal stromal cells from the BM of AML patients (when compared to the BM of healthy people)[26]. Since there have been indications that interactions between HSC and stroma in the niches can influence HSCs[5,27] it is conceivable that elderly AML blasts are more affected by the environment of the abnormal niche.

Also noteworthy, the hypermethylations in *RPTOR* (regulator of MTOR) and *BAIAP2* (regulator of CDC42) implicate mTOR pathway and cell polarity in the distinction of elderly AML for young AML. Both mechanisms have been highlighted as **cell-intrinsic**

mechanisms that contribute to the phenotypes of aged HSCs[3]. This indicated that the altered region of chromosome 17 could be reflecting the origin of elderly AML from HSCs affected by age.

We inspected the list of regions with differential methylation levels in elderly AML for genes with **known associations to age and/or cancer**. Therefore, we searched the literature for corroborations that specific genes could be implicated in AML pathophysiology and/or specific age phenotypes. Lastly, we observed if certain levels of DNA methylation were associated with genes that have previously been **correlated to poor survival in AML**. We detected some DMRs are consistent with other reports which could introduce particular genes as good candidates for further study. However, the majority of DMRs of the elderly AML were not cited in literature or were contrary to expectations.

We found **some candidate genes** in genes that are **related to age**. Due to the lack of correlation of DNA methylation levels to the age of patients with AML (that we have discussed) we expected not many of the genes that distinguish the elderly AML epigenetic profile for the younger AML would have been related to age. We found 7% of the genes in the GenAge database were genes related to DMRs of the elderly cluster (vs the young cluster).

We thought the **differential methylation in *ESR1*** (which encodes the estrogen receptor 1) might be significant since this has been related to age and cancer. It was hypermethylated in the BM of AML patients vs healthy people[137] and was the first shown to become hypermethylated in colon with increasing age[166]. As we already mentioned, to label genes as age or cancer related is not easy since it is not a trivial distinction. Horvath *et al.* summed-up cancer vs age DNA methylation changes in this concise sentence: *"Important cancer-related genes become hypermethylated during aging, including those encoding the estrogen receptor, insulin growth factor, and E-cadherin, and key developmental genes"*[158]*.* Therefore *ESR1* was already a known overlap of DNA methylation changes related to age and cancer.

In the study of Bullinger *et al.*[137] (where *ESR1* was also found hypermethylated in AML) the high levels of *ESR1* methylation among AML patients were found to be a strong poor prognosis predictor[137]. On the contrary, our analysis determined that *ESR1* had a region before the gene coding region with low methylation levels in the elderly group of patients (that accordingly had lower survival probability) and TCGA patients with higher expression of this receptor were the ones with the unfavorable prognosis. The discrepancy is of unclear significance and would merit further investigation.

Therefore, from our age and cancer list, we would emphasize the **differential methylation of a region in *SOCS2*** (which was lower in elderly patients) that was accompanied by the corroboration that **high expression of the gene constitutes a poor prognosis** predictor in the TCGA AML cohort. *SOCS2* expresses a protein connected to the insulin signaling pathway and is a feedback inhibitor of the JAK-STAT cascade[136]. Both pathways are very connected to leukemic diseases, namely AML and ALL. High *SOCS2* expression was shown to be part of the molecular signature of the most primitive HSCs and was linked to AML subsets of unfavorable outcome[94,167]. This was consistent with our results making this **one of the candidates with more potential** for exploration as a prognosis marker.

We could find several genes that other studies found to have **epigenetic marks characteristic of AML** when compared to healthy samples. We should highlight among these were *KIAA1447* and *FSCN2*, genes in the highly methylated region of the tip of chromosome 17. The corroboration with the study of Bullinger *et al.* offered us a substantial suggestion that the many hypermethylations in this region might be **physiologically relevant for AML phenotypes**.

Many **survival markers** have been suggested in AML, several of these were based in DNA methylation levels defining AML groups with poor survival (see Table 1.4.1).
While these studies could not confirm each other's biomarkers of poor survival prognosis, many of these genes were found in our DMR lists. The biggest commonality was with the Marccuci *et al*.[95] study, through 22 genes with epigenetic marks. Most importantly this overlap included 3 (*CD34*, *SCRN1*, *F2RL1*) of the 7 genes score (*CD34*, *RHOC*, *SCRN1*, *F2RL1*, *FAM92A1*, *MIR155HG*, and *VWA8*) identified in their study. Notably, in their study[95], all 7 had both promoter low methylation and high gene expression associated with low overall survival[95].

Therefore, we found several genes with suggested involvement in the elderly AML pathogenesis that could be developed into prognostic markers in AML. The low DNA methylation levels of ***F2RL1, HIVEP3*** **and** ***SOCS2*** **were particularly interesting** because, besides having been found by others, we could confirm their high gene expression was associated with low overall survival in the TCGA cohort.

Collectively the results of the DMR analysis showed many genes could be valuable biomarkers of elderly AML. The prediction value of these differential DNA methylation levels will have to be further evaluated in other cohorts. **Confirmation studies are needed** in order to design potential therapies using these targets to specify treatment strategies to elderly patients.

5.3 Interconnected genetic and epigenetics

We also examined the possible **roles of genetic alterations in the general epigenetic patterns** of AML by observing the integrated genetic and epigenetic profiles.

We found the **cluster defining genetic lesions** in our unsupervised clustering of epigenetic levels in promoters were **similar** to the descriptions in the TCGA report[37]. Most notably, similar groups were perceived using the genomic regions annotated as CpC-sparse regions. In both cases there were groups with similar lesions bearing defined epigenetic profiles, *IDH1/2* mutated, CBF samples, or co-mutated *DNMT3A/NPM1/FLT3*. In our analysis, these groups were possibly more pronounced due to the added samples.

Furthermore, the correlation between genetic lesions and DNA methylation defined groups as seen in Glass *et al.*[91] was **partially comparable to our findings**. Unsupervised clustering of DNA methylation levels in the SAL elderly and TCGA cohorts showed some **analogous molecular groups**. For example, the triple mutated *DNMT3A*/*NPM1*/*FLT3* group they termed *DNMT3A* and the CBF they termed Inv(16). But we could not confirm the existence of an epigenetic group with *NPM1*/*FLT3* (they termed *NPM1*) and there was no segregation of *IDH1* vs *IDH2* mutations into the 2 different groups (that they termed *IDH1*/*DNMT3A* and *IDH2*). Instead, the integrated cohorts (SAL elderly and TCGA) demonstrated the existence of an epigenetic profile that defined a group with ***IDH1/2* mutations combined**. Additionally, we found **other epigenetic groups** like the one with **complex samples**, not found in Glass *et al.*[91]. They missed this possibly due to the composition of their cohort that only included 3% of complex samples while our integrated cohorts (SAL elderly and TCGA) had about 13%. Not surprisingly, they did not report a good separation of young and elderly epigenetic profiles since they had only 12% of patients 60 years or older. Hence, comparisons are limited since some of the differences between the epigenetic classifications found using these cohorts might be due to their fundamentally different age composition.

5.3.1 DNA instability features of elderly AML

Notably, with this extended dataset the epigenetic profiles of the samples from elderly AML patients could be associated to mutations in *IDH1/2* or *TP53* or *IDH*/*DNMT3A* and connected to Group C. This indicated that elderly samples were epigenetically comparable to samples with genetic profiles **suggestive of low DNA stability**. Not only because of the mutations in *DNMT3A* and DNA repair proteins (that we previously mentioned connected to group C) but also because of the many *IDH1* mutations. The latter

were recently shown to affect the expression of ATM and create a situation of low DNA stability through this defect in DNA damage repair[80].

Moreover, the **shores of CpG islands** have revealed good correlations to gene expression in many cancer types. In AML they have produced tight patterns of methylation that were defined by specific genetic alterations as was seen by Glass *et al.*[91] using a new whole-genome approach. Therefore, we decided to analyse the epigenetic profiles of CpG in island shores using the 450,000 CpGs we had available. The profiles revealed similar conclusions to the analysis of promoters but **showed more clearly the epigenetic similarities** of the elderly samples with the samples of complex/*TP53* group, independently of *IDH* mutations.

In addition, some proteins connected to DNA repair were differently affected in the cluster of the elderly, with hypermethylated regions associated to *RAD52, RAD23A, XRCC3, TP63* and *MGMT* and a hypomethylated region related to *TP73*.

All these epigenetic observations matched the assumption drawn from the genetic identity of the elderly AML samples that had already pointed to the **similarities of these samples to samples with impaired DNA repair**. These conjectures highlighted another possible reason for the resistance to treatment and consequently the bad prognosis perceived in the elderly AML.

5.3.2 Methylation profiles within SAL elderly AML

In other AML cohorts distinct methylation patterns have been described for different molecular and cytogenetic groups (including for samples with mutations in *NPM1*, *CEBPA*, *IDH1/2*, *DNMT3A* or *RUNX1*)[89,168], therefore we expected **distinct epigenetic signatures** to exist in the SAL elderly AML. Upon dissection of the DNA methylation profiles of elderly AML we found distinct signatures of samples with **mutations in epigenetic regulators** *IDH1/2* and *DNMT3A*.

These two signatures, with **divergent methylation patterns**, revealed distinct biological functions over-represented among the genes aberrantly methylated. While *DNMT3A* mutations were only associated to differential methylation on members of the HOX family[66], patients with mutations in ***IDHs*** **had a stronger signature**, with DMRs localized in genes of transcription factors that are important in differentiation of the myeloid lineage[79], but also in genes of other biosynthetic and metabolic processes. The restricted amount of DMRs found for samples with *DNMT3A* mutations could be due to effects of the alternation of commutations in *IDH1/2* or in *NPM1*/*FLT3*. Whereas the signature of *IDH1/2* mutations was stronger **despite the split between samples** with or without *DNMT3A* mutations.

Last of all, in the case of *NPM1* and/or *FLT3* mutated samples (vs the respective non-mutated) several DMRs could be found, even though these proteins are not known for being direct influencers of DNA methylation. No inferences were made from this for the cause is unclear since the large majority of these samples also bear mutations in *DNMT3A*.

We concluded, DNA methylation profiles for samples with specific mutations that have been described in younger cohorts using other techniques like HELP[79,89] and ERRBS[91], but also the 450K array[66], were **comparable to the findings in the SAL elderly AML**. The presence of *DNMT3A* mutations had been previously connected to hypomethylations in HOX genes[66], while the *IDH* mutations were previously correlated to many hypermethylations[79].

5.3.3 Specific targets in elderly AML

Finally, we would like to remark, these genetic and methylation analyses underscored the **interconnection of regulatory mechanisms in elderly AML**. Due to the unresponsiveness of elderly AML to intense chemotherapy, these profiles hold important information for the design of more attractive treatment regimens for this group of patients.

(i) Based on the high frequency of mutations in epigenetic regulators in the elderly AML, the use **hypomethylating agents**[101,169] need to be explored more sophisticatedly. This will include studies testing combinational therapies that are currently under accessment[170,171]. On the other hand, patients within the major genetic subgroup, with mutations in chromatin and splicing regulators, should be correlated to treatment response, since these mutations are also observed in MDS where azacitidine and decitabine treatments (demethylation agents) have shown promising results[172,173].

(ii) Due to the high prevalence of mutations in *IDH1/2* and their very particular epigenetic signature, the use of specific **IDH inhibitors**[69,174,175] is of high relevance specifically for elderly AML. This is now possible since the recent approval of Enasidenib[73].

(iii) As several differentiation pathways were strongly affected by hypermethylation, elderly AML patients may benefit from **differentiation therapy**. Interestingly, all-trans-retinoic acid was shown to induce myeloid differentiation in *IDH* mutated AML[176] and might still be an alternative in the elderly.

(iv) Since the elderly samples were genetically and epigenetically correlated to DNA instability perhaps treatment of elderly AML should be adapted accordingly. The possibilities of obtaining synthetic lethality by **targeting DNA damage response pathways** that are underway should be applied to elderly AML (PARP, ATR, ATM, CHK or DNA-PK inhibitors)[177]. The poly(ADP-ribose) polymerase (PARP) inhibitor olaparib has

already shown therapeutic benefits in BRCA-mutant ovarian and breast cancer[178] and induction of apoptosis of AML *in vitro*[179].

Until enough data about specific treatments that would benefit elderly AML can be defined studies like ours should be taken into account since it at least they imply that **not all elderly patients with AML should be treated equal**. While some elderly patients share genetic and epigenetic profiles with younger patients (and would clearly benefit from treatments applied to younger patients), most patients show an elderly AML with genetic and epigenetic reasons for treatment resistance and would **need a different approach**. Therefore, a first step would be to use studies like ours to insert patients into specific groups, defined by **integrated genetic and epigenetic data**, and allow patients not be excluded from treatment only on the bases of age and fitness.

Further advances in treatment will surely be accomplished by confirming and finding additional evidence of the **heterogeneity** and the **specificity** of elderly AML.

CHAPTER 6. CONCLUSION

The main aim of this study was to develop a feasible approach to find the specificity of AML in the elderly that might be contributing to its distinct poor prognosis. We investigated genetic alterations and global epigenetic changes in elderly AML and found both these rigorous determinations were necessary to define a specific molecular profile for elderly AML.

We concluded, elderly AML is a specific entity that harbors specific characteristic genetic and epigenetic patterns, which likely contribute to the treatment resistance in this disease entity.

We found a high frequency of genetic alterations in spliceosome components, epigenetic regulators and in DNA repair factors, the latter being associated with poor prognosis. The molecular categorization of elderly AML into groups according to mutation patterns in DNMT3A/DNA repair or NPM1/RNA processing proteins underscored its distinct biology.

Epigenetic landscapes also separated elderly AML into a particular leukemic group with distinct DNA methylation levels in many gene promoter regions. In general, the disease showed epigenetic similarities to samples of high DNA instability, like samples with complex karyotypes and the samples of a major epigenetic subgroup formed by *IDH1/2* (and/or *DNMT3A* mutants). Methylation levels of regions correlated to elderly AML showed dependencies not only on age but also on two particular genetic groups (*IDH1/2* or *DNMT3A/NPM1/FLT3*). At least an extended region on the tip of the long arm of chromosome 17 could be particularly correlated to poor survival probability.

The CpGs with particular methylation levels for the elderly AML entity overlapped genes known to be involved in age, cancer and particularly AML pathogenesis. Several of them showed potential to be developed into epigenetic-based prognosis markers.

After this last 2 years, with our data and data gathered by others, the field can consider a new view of elderly AML. Agreeing that age is a determinant of elderly AML biology and treatment response because it is connected to specific genetic and epigenetic profiles with its own complexity.

We hope our characterization of genetic and epigenetic patterns of the specific elderly AML group may guide the future development of new strategies to adapt treatments of this unfavorable AML entity.

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APPENDIX A

Table of detailed clinical data for the 93 patients of SAL elderly AML

ID	Material	Age	Sex	Diagnosis	dysplasia	#Blasts	LDH	Leukos	FLT3_ITD	*NPM1c* _ Sanger	NPM1_ NGS	NPM1_ all	Cytogen.	Int_Chem	Response	Surv_ Status	Surv_ Time	#Muts_NGS
S15	BM	65	w	deNovo	1	60	309	2	0	0	0	0	Complex	Y	NA	0	1.3	4
35	BM	66	w	sAML	1	50	161	1.1	0	0	0	0	Others	Y	0	1	0.6	11
36	BM	66	m	deNovo	0	95	1794	60	NA	0	0	0	NA	Y	1	0	59.5	8
48	BM	66	m	deNovo	1	70	168	1.8	0	0	0	0	CN	Y	0	1	17.5	10
59	PB	66	w	deNovo	0	90	1210	126	1	1	1	1	Others	Y	1	0	8.7	6
S32	BM	66	w	deNovo	NA	63.5	812	84.1	0	1	1	1	Others	Y	1	1	16.8	7
S47	BM	66	w	deNovo	NA	75.5	272	11	1	1	1	1	CN	Y	1	1	9.9	7
44	BM	67	w	sAML	NA	NA	168	4.1	NA	0	0	0	NA	Y	1	1	28.7	8
63	BM	67	m	deNovo	0	70	390	16.45	0	0	0	0	CN	Y	1	0	15.1	11
S10	BM	67	m	deNovo	0	95	267	42	0	0	0	0	CBF	Y	1	0	7.5	9
S18	BM	67	w	deNovo	NA	70.5	456	41.4	0	0	0	0	Others	Y	1	1	15.4	10
S33	BM	67	w	t-AML	1	30	489	1.47	0	0	0	0	NA	N	0	0	6.5	4
S34	BM	67	m	deNovo	NA	70.5	505	8.8	0	0	0	0	Others	Y	1	1	14.8	8
S46	BM	67	w	deNovo	NA	82	486	14.6	1	0	0	0	CN	Y	1	0	12.4	9
S61	BM	67	m	deNovo	NA	67.5	1254	82.8	0	0	0	0	CN	Y	0	1	1.7	10
S66	BM	67	w	deNovo	NA	96	930	110.8	1	1	1	1	CN	Y	1	1	16.4	6
24	BM	68	m	deNovo	1	80	405	0.86	1	1	0	1	CN	Y	1	1	12.7	10
34	BM	68	w	deNovo	1	75	274	1.4	NA	0	0	0	CN	Y	1	1	54.0	7
42	BM	68	m	deNovo	1	95	369	27.8	NA	NA	1	1	CN	Y	1	0	91.8	9
62	BM	68	m	sAML	1	80	277	2.2	0	0	0	0	CN	Y	0	0	46.2	9
S19	BM	68	m	deNovo	NA	93.5	144	3.8	0	0	0	0	Others	Y	1	1	16.1	7
S53	BM	68	w	deNovo	NA	70	234	1.3	0	0	0	0	CN	Y	1	0	18.1	7
S59	BM	68	m	deNovo	NA	86	4219	114.2	0	0	0	0	Complex	Y	0	1	6.8	9
S65	BM	68	m	deNovo	NA	76.5	407	55.2	1	1	1	1	CN	Y	1	1	23.9	10
S40	BM	69	w	deNovo	NA	91	537	26	0	0	0	0	CBF	Y	1	1	17.8	8
S48	BM	69	w	deNovo	NA	97.5	330	16.2	0	0	0	0	Complex	Y	1	1	5.4	7
S54	BM	69	w	t-AML	NA	68	482	14.7	0	0	0	0	Others	Y	0	1	2.2	13
10	BM	70	m	deNovo	NA	25	223	1.3	0	0	0	0	CN	Y	1	1	29.5	11
32	BM	70	w	sAML	NA	NA	333	1.76	0	0	0	0	Others	N	0	1	8.5	7
33	BM	70	m	deNovo	0	85	464	5.9	NA	1	1	1	NA	Y	0	1	0.7	9
38	BM	70	m	deNovo	1	70	2015	15	0	0	0	0	Others	Y	1	0	63.7	12
64	BM	70	w	sAML	0	60	461	1.38	0	0	0	0	Complex	Y	0	1	7.8	5
S02	BM	70	m	deNovo	1	95	409	80	0	1	1	1	CN	Y	1	0	5.2	5
S05	BM	70	m	sAML	1	30	518	13.32	NA	0	0	0	Complex	N	0	0	2.1	6
S14	BM	70	m	t-AML	1	25	154	0.91	0	1	0	1	Others	N	0	1	1.9	7
S21	BM	70	w	deNovo	1	99	1077	62.71	1	0	0	0	Others	Y	1	0	20.0	6
S22	BM	70	w	t-AML	NA	64	249	4.4	0	0	0	0	Others	Y	1	1	19.2	13
S45	BM	70	w	deNovo	NA	94	744	81.9	1	1	1	1	Others	Y	1	1	12.9	4
22	BM	71	w	deNovo	1	80	931	47.54	NA	0	0	0	Complex	Y	0	1	1.2	3
37	BM	71	m	deNovo	0	90	300	1	NA	0	0	0	Others	Y	1	1	10.1	9
55	BM	71	m	deNovo	1	65	254	4.4	NA	0	0	0	Complex	N	0	0	0.2	9
S26	BM	71	m	deNovo	1	40	138	1.9	0	0	0	0	Others	Y	0	0	11.0	6
S27	BM	71	w	deNovo	0	70	170	1.8	NA	0	0	0	CN	Y	1	1	19.4	5
S37	BM	71	m	deNovo	NA	80	199	1.8	0	0	0	0	CN	Y	1	1	20.0	10
43	BM	72	m	sAML	1	85	346	35.7	NA	0	0	0	NA	Y	1	1	16.9	7
47	BM	72	w	deNovo	0	95	553	20	NA	0	0	0	Complex	Y	0	0	1.3	11
S13	BM	72	m	deNovo	1	30	190	1.49	0	1	0	1	CN	Y	1	0	32.3	1
S16	BM	72	w	sAML	1	50	388	2.53	NA	0	0	0	NA	N	0	0	3.8	3
S35	BM	72	m	deNovo	NA	66	367	6.6	0	0	0	0	Others	Y	1	1	38.6	8
S43	BM	72	w	deNovo	NA	87.5	636	71.9	0	0	0	0	Others	Y	1	1	25.8	9
S58	BM	72	w	sAML	NA	69.5	218	4.7	0	0	0	0	CN	Y	0	1	9.2	7
8	BM	73	w	deNovo	0	80	319	2.15	0	0	0	0	Complex	N	0	1	0.8	6
11	BM	73	w	deNovo	0	99	498	69	1	0	0	0	CN	Y	0	1	3.9	6
S12	BM	73	m	sAML	NA	97	561	47	0	0	0	0	NA	Y	0	1	5.2	6
S39	BM	73	w	deNovo	NA	85.5	1482	10.4	0	0	0	0	CBF	Y	1	0	2.7	4
S57	BM	73	m	sAML	NA	72.5	996	60.7	1	1	1	1	Others	Y	0	1	6.2	5
S64	BM	73	w	sAML	NA	69	276	47	0	0	0	0	Others	Y	0	1	3.0	6
1	BM	74	m	t-AML	1	95	189	1.68	0	0	0	0	Complex	N	0	0	2.2	23
23	BM	74	m	deNovo	0	80	582	56.45	1	1	0	1	CN	Y	1	1	15.5	4
S03	BM	74	m	sAML	1	90	1992	195.76	0	0	0	0	CN	Y	0	1	6.4	9
S17	BM	74	m	deNovo	0	60	333	42.5	0	0	0	0	Others	Y	1	1	27.7	7
S62	BM	74	w	deNovo	NA	88.5	882	160.8	1	0	0	0	Others	Y	0	1	2.6	6
S63	BM	74	m	deNovo	NA	64.5	536	1.4	0	0	0	0	Others	Y	1	1	12.7	4
6	BM	75	m	sAML	0	95	542	91	0	0	0	0	Others	Y	1	1	13.0	10
7	BM	75	m	deNovo	0	99	509	189	0	1	1	1	CN	Y	1	0	15.7	10
S30	BM	75	w	deNovo	NA	70.5	328	10.2	0	0	0	0	Others	Y	1	1	21.9	7
S38	BM	75	w	deNovo	NA	70.5	169	1	0	0	0	0	CN	Y	1	0	6.9	7
S51	BM	75	m	deNovo	NA	95	1184	36	0	0	0	0	Others	Y	0	1	14.8	11
12	BM	76	m	deNovo	1	90	604	149	0	0	0	0	CN	Y	0	1	13.2	12
S06	BM	77	m	deNovo	1	90	431	85	1	1	1	1	NA	N	0	0	5.5	6
S44	BM	77	m	deNovo	NA	63	265	14.9	0	0	0	0	CBF	Y	1	1	20.2	6
20	BM	78	m	deNovo	1	45	363	1.75	0	0	0	0	Others	N	0	1	0.2	4
46	BM	78	m	deNovo	0	95	462	33.8	NA	1	1	1	CN	Y	1	1	63.5	9
S20	BM	78	w	deNovo	NA	64	552	18	0	0	0	0	CBF	Y	1	0	46.9	7
S36	BM	78	w	deNovo	NA	74	714	209.6	1	0	0	0	CN	Y	1	1	7.3	7
S42	BM	78	m	sAML	NA	72.5	516	8.7	1	0	0	0	Others	Y	1	1	43.3	7
58	BM	79	w	deNovo	1	75	647	16	NA	0	0	0	Others	N	0	0	2.1	6
S29	BM	79	m	deNovo	NA	NA	239	18.58	0	0	0	0	NA	N	0	0	0.1	8
S52	BM	79	w	deNovo	NA	64.5	175	1.1	0	1	1	1	Others	Y	1	0	66.0	6
S56	BM	79	m	deNovo	NA	65	392	6.2	0	0	0	0	Others	Y	0	1	3.9	13
30	BM	80	w	deNovo	1	40	251	1.19	0	0	0	0	NA	N	0	0	1.3	6
S23	BM	80	m	deNovo	NA	NA	279	3.7	NA	0	0	0	NA	Y	0	1	1.1	3
S55	BM	80	m	sAML	NA	64.5	454	4.4	0	0	0	0	Complex	Y	0	1	3.1	5
S60	BM	80	w	deNovo	NA	68	1110	45.5	0	0	0	0	Complex	Y	0	1	2.0	3
19	BM	81	m	deNovo	1	95	3340	178.58	NA	1	1	1	NA	NA	NA	1	0.9	12
21	BM	81	w	deNovo	NA	40	1094	82.5	0	0	0	0	CN	N	0	1	2.3	12
31	BM	81	w	deNovo	0	80	398	14.8	NA	1	0	1	CN	N	0	1	9.7	11
26	BM	82	w	deNovo	1	50	235	5	NA	0	0	0	NA	N	0	1	7.6	6
29	BM	83	w	deNovo	NA	NA	NA	NA	NA	0	0	0	NA	NA	NA	NA	NA	3
40	BM	83	m	sAML	0	55	378	4.38	NA	0	0	0	NA	N	0	0	8.0	6
25	BM	86	w	deNovo	0	95	386	63.3	0	0	0	0	NA	N	0	1	0.0	12
28	BM	88	w	deNovo	0	80	263	3.33	NA	0	0	0	NA	N	0	0	0.4	5
S25	BM	90	w	sAML	1	75	1712	127.5	NA	0	0	0	NA	NA	NA	1	0.1	12

APPENDIX B

Table of information from the genetic alterations in the SAL elderly AML

ID	Type Mut	Chrom	Position	Ref Allele	VarAllele	Total Reads	Ref Reads	Var Reads	VarFreq	GeneName	GeneID	Effect	HGVSc	HGVSp	EXON	CANONICAL	GMAF	Existing_variation
19	INDEL	chr9	133759780	C	-GAG	330	186	144	0.434	ABL1	ENSG00000097007	CODON_CHANGE_PLUS_CODON_DELETION	ENST00000372348.2:c.216 1delG	ENSP00000361423.2:p.Glu72ArgfsTer74	11/11	YES	-	-
19	INDEL	chr9	133759780	C	-GAG	330	186	144	0.434	ABL1	ENSG00000097007	CODON_CHANGE_PLUS_CODON_DELETION	ENST00000318560.5:c.2104delG	ENSP00000323315.5:p.Glu702ArgfsTer74	11/11	-	-	-
S21	SNV	chr9	133760165	C	T	285	159	126	0.442	ABL1	ENSG00000097007	NON_SYNONYMOUS_CODING	ENST00000318560.5:c.2488C>T	ENSP00000323315.5:p.Leu830Phe	11/11	-	-	-
S21	SNV	chr9	133760165	C	T	285	159	126	0.442	ABL1	ENSG00000097007	NON_SYNONYMOUS_CODING	ENST00000372348.2:c.2545C>T	ENSP00000361423.2:p.Leu849Phe	11/11	YES	-	-
62	SNV	chr19	6333484	C	T	189	98	91	0.472	ACER1	ENSG00000167769	NON_SYNONYMOUS_CODING	ENST00000301452.4:c.79G>A	ENSP00000301452.3:p.Glu27Lys	1/6	YES	-	-
S40	SNV	chr2	237489592	C	T	784	383	401	0.51	ACKR3	ENSG00000144476	NON_SYNONYMOUS_CODING	ENST00000272928.3:c.484C>T	ENSP00000272928.3:p.Arg162Cys	2/2	YES	-	-
S40	SNV	chr2	237489592	C	T	784	383	401	0.51	ACKR3	ENSG00000144476	NON_SYNONYMOUS_CODING	ENST00000447924.1:c.484C>T	ENSP00000405945.1:p.Arg162Cys	2/2	-	-	-
1	SNV	chr2	223806250	C	T	160	86	74	0.462	ACSL3	ENSG00000123983	NON_SYNONYMOUS_CODING	ENST00000357430.3:c.204 1C>T	ENSP00000350012.3:p.Arg68 1Cys	17/17	YES	-	COSM210156,COSM231696
1	SNV	chr2	223806250	C	T	160	86	74	0.462	ACSL3	ENSG00000123983	NON_SYNONYMOUS_CODING	ENST00000392066.3:c.204 1C>T	ENSP00000375918.3:p.Arg68 1Cys	16/16	-	-	COSM210156,COSM231696
19	SNV	chr5	131324260	G	T	310	165	145	0.468	ACSL6	ENSG00000164398	NON_SYNONYMOUS_CODING	ENST00000379272.2:c.611C>A	ENSP00000368574.2:p.Ala204Asp	7/22	-	-	-
19	SNV	chr5	131324260	G	T	310	165	145	0.468	ACSL6	ENSG00000164398	NON_SYNONYMOUS_CODING	ENST00000431707.1:c.506C>A	ENSP00000413329.1:p.Ala169Asp	6/21	-	-	-
21	SNV	chr2	100623753	T	C	299	141	158	0.528	AFF3	ENSG00000144218	NON_SYNONYMOUS_CODING	ENST00000423966.1:c.344A>G	ENSP00000396582.1:p.Asp115Gly	5/5	-	-	-
21	SNV	chr2	100623753	T	C	299	141	158	0.528	AFF3	ENSG00000144218	NON_SYNONYMOUS_CODING	ENST00000409579.1:c.419A>G	ENSP00000386834.1:p.Asp140Gly	6/25	-	-	-
21	SNV	chr2	100623753	T	C	299	141	158	0.528	AFF3	ENSG00000144218	NON_SYNONYMOUS_CODING	ENST00000317233.4:c.344A>G	ENSP00000317421.4:p.Asp115Gly	5/24	-	-	-
21	SNV	chr2	100623753	T	C	299	141	158	0.528	AFF3	ENSG00000144218	NON_SYNONYMOUS_CODING	ENST00000441400.1:c.344A>G	ENSP00000399795.1:p.Asp115Gly	4/4	-	-	-
21	SNV	chr2	100623753	T	C	299	141	158	0.528	AFF3	ENSG00000144218	NON_SYNONYMOUS_CODING	ENST00000432037.1:c.344A>G	ENSP00000406484.1:p.Asp115Gly	3/3	-	-	-
21	SNV	chr2	100623753	T	C	299	141	158	0.528	AFF3	ENSG00000144218	NON_SYNONYMOUS_CODING	ENST00000356421.2:c.419A>G	ENSP00000348793.2:p.Asp140Gly	5/24	YES	-	-
21	SNV	chr2	100623753	T	C	299	141	158	0.528	AFF3	ENSG00000144218	NON_SYNONYMOUS_CODING	ENST00000409236.2:c.344A>G	ENSP00000387207.1:p.Asp115Gly	4/23	-	-	-
24	SNV	chr7	91726229	A	G	563	302	261	0.464	AKAP9	ENSG00000127914	NON_SYNONYMOUS_CODING	ENST00000359028.2:c.9968A>G	ENSP00000351922.2:p.Glu3323Gly	41/50	-	-	-
24	SNV	chr7	91726229	A	G	563	302	261	0.464	AKAP9	ENSG00000127914	NON_SYNONYMOUS_CODING	ENST00000356239.3:c.9956A>G	ENSP00000348573.3:p.Glu3319Gly	41/50	YES	-	-
24	SNV	chr7	91726229	A	G	563	302	261	0.464	AKAP9	ENSG00000127914	NON_SYNONYMOUS_CODING	ENST00000358100.2:c.9806A>G	ENSP00000350813.2:p.Glu3269Gly	40/49	-	-	-
24	SNV	chr7	91726229	A	G	563	302	261	0.464	AKAP9	ENSG00000127914	NON_SYNONYMOUS_CODING	ENST00000394534.2:c.9494A>G	ENSP00000378042.2:p.Glu1165Gly	15/23	-	-	-
S10	SNV	chr7	91709362	A	G	515	287	228	0.443	AKAP9	ENSG00000127914	NON_SYNONYMOUS_CODING	ENST00000358100.2:c.7951A>G	ENSP00000350813.2:p.Lys2651Glu	32/49	-	-	-
S10	SNV	chr7	91709362	A	G	515	287	228	0.443	AKAP9	ENSG00000127914	NON_SYNONYMOUS_CODING	ENST00000359028.2:c.7951A>G	ENSP00000351922.2:p.Lys2651Glu	32/50	-	-	-
S10	SNV	chr7	91709362	A	G	515	287	228	0.443	AKAP9	ENSG00000127914	NON_SYNONYMOUS_CODING	ENST00000356239.3:c.7915A>G	ENSP00000348573.3:p.Lys2639Glu	31/50	YES	-	-
S10	SNV	chr7	91709362	A	G	515	287	228	0.443	AKAP9	ENSG00000127914	NON_SYNONYMOUS_CODING	ENST00000394534.2:c.1453A>G	ENSP00000378042.2:p.Lys485Glu	5/23	-	-	-
S42	SNV	chr7	91718853	A	T	475	225	250	0.526	AKAP9	ENSG00000127914	NON_SYNONYMOUS_CODING	ENST00000435423.1:c.802A>T	ENSP00000406937.1:p.Tyr268Phe	6/6	-	0.0030	rs180926926
S48	SNV	chr7	91631344	T	G	339	178	161	0.475	AKAP9	ENSG00000127914	NON_SYNONYMOUS_CODING	ENST00000358100.2:c.2149T>G	ENSP00000350813.2:p.Leu717Val	9/49	-	-	-
S48	SNV	chr7	91631344	T	G	339	178	161	0.475	AKAP9	ENSG00000127914	NON_SYNONYMOUS_CODING	ENST00000359028.2:c.2149T>G	ENSP00000351922.2:p.Leu717Val	9/50	-	-	-
S48	SNV	chr7	91631344	T	G	339	178	161	0.475	AKAP9	ENSG00000127914	NON_SYNONYMOUS_CODING	ENST00000356239.3:c.2113T>G	ENSP00000348573.3:p.Leu705Val	8/50	YES	-	-
S44	SNV	chr14	105258971	C	T	630	311	319	0.506	AKT1	ENSG00000142208	NON_SYNONYMOUS_CODING	ENST00000407796.2:c.10G>A	ENSP00000384293.2:p.Val4Met	2/14	-	-	COSM3814333
S44	SNV	chr14	105258971	C	T	630	311	319	0.506	AKT1	ENSG00000142208	NON_SYNONYMOUS_CODING	ENST00000555528.1:c.10G>A	ENSP00000450688.1:p.Val4Met	2/14	-	-	COSM3814333
S44	SNV	chr14	105258971	C	T	630	311	319	0.506	AKT1	ENSG00000142208	NON_SYNONYMOUS_CODING	ENST00000554581.1:c.10G>A	ENSP00000451828.1:p.Val4Met	1/13	YES	-	COSM3814333
S44	SNV	chr14	105258971	C	T	630	311	319	0.506	AKT1	ENSG00000142208	NON_SYNONYMOUS_CODING	ENST00000402615.2:c.10G>A	ENSP00000385326.2:p.Val4Met	2/14	-	-	COSM3814333
S44	SNV	chr14	105258971	C	T	630	311	319	0.506	AKT1	ENSG00000142208	NON_SYNONYMOUS_CODING	ENST00000554848.1:c.10G>A	ENSP00000451166.1:p.Val4Met	2/14	-	-	COSM3814333
S44	SNV	chr14	105258971	C	T	630	311	319	0.506	AKT1	ENSG00000142208	NON_SYNONYMOUS_CODING	ENST00000555926.1:c.10G>A	ENSP00000451824.1:p.Val4Met	3/5	-	-	COSM3814333
S44	SNV	chr14	105258971	C	T	630	311	319	0.506	AKT1	ENSG00000142208	NON_SYNONYMOUS_CODING	ENST00000349310.3:c.10G>A	ENSP00000267020:p.Val4Met	3/15	-	-	COSM3814333
48	SNV	chr19	4076214	C	T	604	301	303	0.502	AKT2	ENSG00000105221	NON_SYNONYMOUS_CODING	ENST00000427375.1:c.94G>A	ENSP00000403890.1:p.Asp32Asn	4/5	-	-	-
48	SNV	chr19	4076214	C	T	604	301	303	0.502	AKT2	ENSG00000105221	NON_SYNONYMOUS_CODING	ENST00000392037.1:c.94G>A	ENSP00000375891.1:p.Asp32Asn	2/4	-	-	-
48	SNV	chr19	4076214	C	T	604	301	303	0.502	AKT2	ENSG00000105221	NON_SYNONYMOUS_CODING	ENST00000452077.1:c.94G>A	ENSP00000404083.1:p.Asp32Asn	3/5	-	-	-
48	SNV	chr19	4076214	C	T	604	301	303	0.502	AKT2	ENSG00000105221	NON_SYNONYMOUS_CODING	ENST00000416994.2:c.94G>A	ENSP00000392458.2:p.Asp32Asn	3/4	-	-	-
48	SNV	chr19	4076214	C	T	604	301	303	0.502	AKT2	ENSG00000105221	NON_SYNONYMOUS_CODING	ENST00000456441.1:c.94G>A	ENSP00000396532.1:p.Asp32Asn	4/6	-	-	-
48	SNV	chr19	4076214	C	T	604	301	303	0.502	AKT2	ENSG00000105221	NON_SYNONYMOUS_CODING	ENST00000441941.2:c.94G>A	ENSP00000396968.2:p.Asp32Asn	3/6	-	-	-
48	SNV	chr19	4076214	C	T	604	301	303	0.502	AKT2	ENSG00000105221	NON_SYNONYMOUS_CODING	ENST00000416362.1:c.94G>A	ENSP00000407999.1:p.Asp32Asn	5/7	-	-	-
48	SNV	chr19	4076214	C	T	604	301	303	0.502	AKT2	ENSG00000105221	NON_SYNONYMOUS_CODING	ENST00000392038.2:c.94G>A	ENSP00000375892.2:p.Asp32Asn	3/14	YES	-	-
48	SNV	chr19	4076214	C	T	604	301	303	0.502	AKT2	ENSG00000105221	NON_SYNONYMOUS_CODING	ENST00000423127.1:c.94G>A	ENSP00000403842.1:p.Asp32Asn	5/7	-	-	-
48	SNV	chr19	4076214	C	T	604	301	303	0.502	AKT2	ENSG00000105221	NON_SYNONYMOUS_CODING	ENST00000358335.5:c.94G>A	ENSP00000351095.5:p.Asp32Asn	3/5	-	-	-
48	SNV	chr19	4076214	C	T	604	301	303	0.502	AKT2	ENSG00000105221	NON_SYNONYMOUS_CODING	ENST00000311278.6:c.94G>A	ENSP00000309428.6:p.Asp32Asn	2/12	-	-	-
48	SNV	chr19	4076214	C	T	604	301	303	0.502	AKT2	ENSG00000105221	NON_SYNONYMOUS_CODING	ENST00000424901.1:c.94G>A	ENSP00000399532.1:p.Asp32Asn	2/13	-	-	-
1	SNV	chr12	112204710	G	C	63	46	17	0.27	ALDH2	ENSG00000111275	UTR_5_PRIME	ENST00000416293.3:c.-77G>C	-	1/12	-	-	-
S56	SNV	chr12	112241747	C	G	585	300	285	0.487	ALDH2	ENSG00000111275	STOP_GAINED	ENST00000416293.3:c.1350C>G	ENSP00000403349.3:p.Tyr450Ter	11/12	-	0.0002	rs201939387
S56	SNV	chr12	112241747	C	G	585	300	285	0.487	ALDH2	ENSG00000111275	STOP_GAINED	ENST00000261733.2:c.1491C>G	ENSP00000261733.2:p.Tyr497Ter	12/13	YES	0.0002	rs201939387
S40	SNV	chrX	63410678	T	C	709	337	372	0.525	AMER1	ENSG00000184675	NON_SYNONYMOUS_CODING	ENST00000330258.3:c.2489A>G	ENSP00000329117.3:p.Asn830Ser	2/2	YES	-	-
1	SNV	chr5	112179828	G	C	188	94	94	0.5	APC	ENSG00000134982	UTR_3_PRIME	ENST00000508376.2:c.*5G>C	-	17/17	-	-	-
1	SNV	chr5	112179828	G	C	188	94	94	0.5	APC	ENSG00000134982	UTR_3_PRIME	ENST00000257430.4:c.*5G>C	-	16/16	-	-	-
1	SNV	chr5	112179828	G	C	188	94	94	0.5	APC	ENSG00000134982	UTR_3_PRIME	ENST00000457016.1:c.*5G>C	-	16/16	YES	-	-
S22	SNV	chr5	112164631	G	A	610	318	292	0.479	APC	ENSG00000134982	NON_SYNONYMOUS_CODING	ENST00000457016.1:c.1705G>A	ENSP00000413133.1:p.Val569Met	14/16	YES	-	-
S22	SNV	chr5	112164631	G	A	610	318	292	0.479	APC	ENSG00000134982	NON_SYNONYMOUS_CODING	ENST00000257430.4:c.1705G>A	ENSP00000257430.4:p.Val569Met	14/16	-	-	-
S22	SNV	chr5	112164631	G	A	610	318	292	0.479	APC	ENSG00000134982	NON_SYNONYMOUS_CODING	ENST00000504915.2:c.394G>A	ENSP00000473393.1:p.Val132Met	5/7	-	-	-
S22	SNV	chr5	112164631	G	A	610	318	292	0.479	APC	ENSG00000134982	NON_SYNONYMOUS_CODING	ENST00000508376.2:c.1705G>A	ENSP00000427089.2:p.Val569Met	15/17	-	-	-
S22	SNV	chr5	112164631	G	A	610	318	292	0.479	APC	ENSG00000134982	NON_SYNONYMOUS_CODING	ENST00000507379.1:c.1651G>A	ENSP00000423224.1:p.Val551Met	12/14	-	-	-
S22	SNV	chr5	112164631	G	A	610	318	292	0.479	APC	ENSG00000134982	NON_SYNONYMOUS_CODING	ENST00000512211.2:c.1705G>A	ENSP00000423828.2:p.Val569Met	14/16	-	-	-
S56	SNV	chr5	112175198	C	A	900	467	433	0.481	APC	ENSG00000134982	NON_SYNONYMOUS_CODING	ENST00000508376.2:c.3907C>A	ENSP00000427089.2:p.Gln1303Lys	17/17	-	-	COSM13728,COSM994441
S56	SNV	chr5	112175198	C	A	900	467	433	0.481	APC	ENSG00000134982	NON_SYNONYMOUS_CODING	ENST00000512211.2:c.3907C>A	ENSP00000423828.2:p.Gln1303Lys	16/16	-	-	COSM13728,COSM994441
S56	SNV	chr5	112175198	C	A	900	467	433	0.481	APC	ENSG00000134982	NON_SYNONYMOUS_CODING	ENST00000257430.4:c.3907C>A	ENSP00000257430.4:p.Gln1303Lys	16/16	-	-	COSM13728,COSM994441
S56	SNV	chr5	112175198	C	A	900	467	433	0.481	APC	ENSG00000134982	NON_SYNONYMOUS_CODING	ENST00000457016.1:c.3907C>A	ENSP00000413133.1:p.Gln1303Lys	16/16	YES	-	COSM13728,COSM994441
S58	SNV	chr5	112043547	C	T	483	256	227	0.47	APC	ENSG00000134982	START_GAINED	ENST00000457016.1:c.-51C>T	-	1/16	YES	-	-
S58	SNV	chr5	112043547	C	T	483	256	227	0.47	APC	ENSG00000134982	NON_SYNONYMOUS_CODING	ENST00000507379.1:c.133C>T	ENSP00000423224.1:p.Arg45Cys	1/14	-	-	-
19	SNV	chr2	21252560	A	G	691	373	318	0.46	APOB	ENSG00000084674	NON_SYNONYMOUS_CODING	ENST00000399256.4:c.1568T>C	ENSP00000382200.4:p.Ile523Thr	12/17	-	-	-
19	SNV	chr2	21252560	A	G	691	373	318	0.46	APOB	ENSG00000084674	NON_SYNONYMOUS_CODING	ENST00000233242.1:c.1568T>C	ENSP00000233242.1:p.Ile523Thr	12/29	YES	-	-
21	SNV	chr2	21231788	G	A	380	48	332	0.874	APOB	ENSG00000084674	NON_SYNONYMOUS_CODING	ENST00000233242.1:c.7952G>T	ENSP00000233242.1:p.Thr2651Ile	26/29	YES	-	-
8	SNV	chr2	21247703	T	C	453	248	205	0.452	APOB	ENSG00000084674	NON_SYNONYMOUS_CODING	ENST00000399256.4:c.2450A>G	ENSP00000382200.4:p.Asp817Gly	17/17	-	-	rs576999094
S46	SNV	chr2	21231882	G	A	596	350	246	0.413	APOB	ENSG00000084674	NON_SYNONYMOUS_CODING	ENST00000233242.1:c.7858C>T	ENSP00000233242.1:p.Pro2620Ser	26/29	YES	-	-
S56	SNV	chr2	21260009	C	A	1107	547	560	0.505	APOB	ENSG00000084674	NON_SYNONYMOUS_CODING	ENST00000399256.4:c.656G>T	ENSP00000382200.4:p.Arg219Leu	6/17	-	0.0004	rs201606169
S56	SNV	chr2	21260009	C	A	1107	547	560	0.505	APOB	ENSG00000084674	NON_SYNONYMOUS_CODING	ENST00000233242.1:c.656G>T	ENSP00000233242.1:p.Arg219Leu	6/29	YES	0.0004	rs201606169
55	SNV	chr5	142586795	C	T	414	220	194	0.469	ARHGAP26	ENSG00000145819	NON_SYNONYMOUS_CODING	ENST00000378004.3:c.2021C>T	ENSP00000367243.3:p.Ser674Leu	21/23	-	-	-
55	SNV	chr5	142586795	C	T	414	220	194	0.469	ARHGAP26	ENSG00000145819	NON_SYNONYMOUS_CODING	ENST00000274498.4:c.2021C>T	ENSP00000274498.4:p.Ser674Leu	21/23	YES	-	-
S21	SNV	chr5	142252982	C	T	290	152	138	0.476	ARHGAP26	ENSG00000145819	NON_SYNONYMOUS_CODING	ENST00000378004.3:c.172C>T	ENSP00000367243.3:p.Arg58Trp	2/23	-	-	-
S21	SNV	chr5	142252982	C	T	290	152	138	0.476	ARHGAP26	ENSG00000145819	NON_SYNONYMOUS_CODING	ENST00000274498.4:c.172C>T	ENSP00000274498.4:p.Arg58Trp	2/23	YES	-	-
S21	SNV	chr5	142252982	C	T	290	152	138	0.476	ARHGAP26	ENSG00000145819	NON_SYNONYMOUS_CODING	ENST00000378013.2:c.88C>T	ENSP00000367252.2:p.Arg30Trp	2/6	-	-	-
23	INDEL	chr11	73022097	A	GGGGCCTGGCACCC T	360	245	115	0.319	ARHGEF17	ENSG00000110237	CODON_CHANGE_PLUS_CODON_DELETION	ENST00000263674.3:c.2415delG	ENSP00000263674.3:p.Pro807LeufsTer22	1/21	YES	-	-
44	SNV	chr11	73020773	G	T	614	307	307	0.5	ARHGEF17	ENSG00000110237	NON_SYNONYMOUS_CODING	ENST00000263674.3:c.1090G>T	ENSP00000263674.3:p.Asp364Tyr	1/21	YES	-	-
S54	SNV	chr11	73019814	G	A	152	80	72	0.474	ARHGEF17	ENSG00000110237	NON_SYNONYMOUS_CODING	ENST00000263674.3:c.131G>A	ENSP00000263674.3:p.Cys44Tyr	1/21	YES	-	COSM3687633
31	SNV	chr1	27056304	A	C	153	83	70	0.458	ARID1A	ENSG00000117713	NON_SYNONYMOUS_CODING	ENST00000457599.2:c.1300A>C	ENSP00000387636.2:p.Met434Leu	2/20	-	-	rs1133451
31	SNV	chr1	27056304	A	C	153	83	70	0.458	ARID1A	ENSG00000117713	NON_SYNONYMOUS_CODING	ENST00000374152.2:c.151A>C	ENSP00000363267.2:p.Met51Leu	1/19	-	-	rs1133451
31	SNV	chr1	27056304	A	C	153	83	70	0.458	ARID1A	ENSG00000117713	NON_SYNONYMOUS_CODING	ENST00000324856.7:c.1300A>C	ENSP00000320485.7:p.Met434Leu	2/20	YES	-	rs1133451
31	SNV	chr1	27056304	A	C	153	83	70	0.458	ARID1A	ENSG00000117713	NON_SYNONYMOUS_CODING	ENST00000524572.1:c.151A>C	ENSP00000432473.1:p.Met51Leu	2/3	-	-	rs1133451
35	SNV	chr1	27101699	A	G	294	136	158	0.537	ARID1A	ENSG00000117713	NON_SYNONYMOUS_CODING	ENST00000324856.7:c.4981A>G	ENSP00000320485.7:p.Met1661Val	18/20	YES	-	-
35	SNV	chr1	27101699	A	G	294	136	158	0.537	ARID1A	ENSG00000117713	NON_SYNONYMOUS_CODING	ENST00000457599.2:c.4330A>G	ENSP00000387636.2:p.Met1444Val	18/20	-	-	-
35	SNV	chr1	27101699	A	G	294	136	158	0.537	ARID1A	ENSG00000117713	NON_SYNONYMOUS_CODING	ENST00000374152.2:c.3832A>G	ENSP00000363267.2:p.Met1278Val	17/19	-	-	-
S16	INDEL	chr1	27023620	C	+GCG	200	116	84	0.42	ARID1A	ENSG00000117713	CODON_CHANGE_PLUS_CODON_INSERTION	-	-	1/20	-	-	-
S16	INDEL	chr1	27023620	C	+GCG	200	116	84	0.42	ARID1A	ENSG00000117713	CODON_CHANGE_PLUS_CODON_INSERTION	-	-	1/20	YES	-	-
S26	SNV	chr1	27023402	C	T	131	67	64	0.488	ARID1A	ENSG00000117713	NON_SYNONYMOUS_CODING	ENST00000457599.2:c.508C>T	ENSP00000387636.2:p.His170Tyr	1/20	-	-	-
S26	SNV	chr1	27023402	C	T	131	67	64	0.488	ARID1A	ENSG00000117713	NON_SYNONYMOUS_CODING	ENST00000324856.7:c.508C>T	ENSP00000320485.7:p.His170Tyr	1/20	YES	-	-
S40	SNV	chr1	27023366	C	T	156	75	81	0.519	ARID1A	ENSG00000117713	NON_SYNONYMOUS_CODING	ENST00000324856.7:c.472C>T	ENSP00000320485.7:p.Pro158Ser	1/20	YES	0.0004	rs567246585

S40	SNV	chr1	27023366	C	T	156	75	81	0.519	ARID1A	ENSG00000117713	NON_SYNONYMOUS_CODING	ENST00000457599.2:c.472C>T	ENSP00000387636.2:p.Pro158Ser	1/20	-	0.0004	rs567246585
34	SNV	chr12	46245235	G	A	236	113	123	0.521	ARID2	ENSG00000189079	NON_SYNONYMOUS_CODING	ENST00000422737.1:c.2882G>A	ENSP00000415650.1:p.Gly961Glu	15/20	-	-	-
34	SNV	chr12	46245235	G	A	236	113	123	0.521	ARID2	ENSG00000189079	NON_SYNONYMOUS_CODING	ENST00000444670.1:c.2159G>A	ENSP00000397307.1:p.Gly720Glu	7/13	-	-	-
34	SNV	chr12	46245235	G	A	236	113	123	0.521	ARID2	ENSG00000189079	NON_SYNONYMOUS_CODING	ENST00000334344.6:c.3329G>A	ENSP00000335044.6:p.Gly1110Glu	15/21	YES	-	-
S14	SNV	chr12	46123904	T	G	188	115	73	0.388	ARID2	ENSG00000189079	STOP_GAINED	ENST00000334344.6:c.170T>G	ENSP00000335044.6:p.Leu57Ter	2/21	YES	-	-
S14	SNV	chr12	46123904	T	G	188	115	73	0.388	ARID2	ENSG00000189079	START_GAINED	ENST00000422737.1:c.-278T>G	-	2/20	-	-	-
S51	SNV	chr17	79941449	G	A	347	164	183	0.527	ASPSCR1	ENSG00000169696	NON_SYNONYMOUS_CODING	ENST00000581647.1:c.178G>A	ENSP00000464408.1:p.Asp60Asn	3/5	-	-	-
S51	SNV	chr17	79941449	G	A	347	164	183	0.527	ASPSCR1	ENSG00000169696	NON_SYNONYMOUS_CODING	ENST00000306729.7:c.178G>A	ENSP00000306625.7:p.Asp60Asn	3/17	YES	-	-
S51	SNV	chr17	79941449	G	A	347	164	183	0.527	ASPSCR1	ENSG00000169696	NON_SYNONYMOUS_CODING	ENST00000581484.1:c.169G>A	ENSP00000463316.1:p.Asp57Asn	3/7	-	-	-
S51	SNV	chr17	79941449	G	A	347	164	183	0.527	ASPSCR1	ENSG00000169696	NON_SYNONYMOUS_CODING	ENST00000306739.4:c.178G>A	ENSP00000302176.4:p.Asp60Asn	3/16	-	-	-
S51	SNV	chr17	79941449	G	A	347	164	183	0.527	ASPSCR1	ENSG00000169696	NON_SYNONYMOUS_CODING	ENST00000579684.1:c.120G>A	ENSP00000461984.1:p.Asp41Asn	3/6	-	-	-
S59	SNV	chr17	79935467	C	T	46	26	20	0.435	ASPSCR1	ENSG00000169696	START_GAINED	ENST00000306739.4:c.-56C>T	-	1/16	-	-	-
S59	SNV	chr17	79935467	C	T	46	26	20	0.435	ASPSCR1	ENSG00000169696	START_GAINED	ENST00000306729.7:c.-56C>T	-	1/17	YES	-	-
10	INDEL	chr20	31022402	T	- CACCACTGCCATAGA GAGGCGGC	253	161	92	0.362	ASXL1	ENSG00000171456	FRAME_SHIFT	ENST00000306058.5:c.1873delC	ENSP00000305119.5:p.His625ThrfsTer73	12/12	-	-	-
10	INDEL	chr20	31022402	T	- CACCACTGCCATAGA GAGGCGGC	253	161	92	0.362	ASXL1	ENSG00000171456	FRAME_SHIFT	ENST00000375687.4:c.1888delC	ENSP00000364839.4:p.His630ThrfsTer73	13/13	YES	-	-
12	INDEL	chr20	31022441	A	+G	153	111	42	0.268	ASXL1	ENSG00000171456	FRAME_SHIFT	-	-	13/13	YES	-	COSM1180918,TMP_ESP_20_31022442_31022442
12	INDEL	chr20	31022441	A	+G	153	111	42	0.268	ASXL1	ENSG00000171456	FRAME_SHIFT	-	-	12/12	-	-	COSM1180918,TMP_ESP_20_31022442_31022442
25	INDEL	chr20	31022441	A	+G	145	91	54	0.362	ASXL1	ENSG00000171456	FRAME_SHIFT	-	-	13/13	YES	-	COSM1180918,TMP_ESP_20_31022442_31022442
25	INDEL	chr20	31022441	A	+G	145	91	54	0.362	ASXL1	ENSG00000171456	FRAME_SHIFT	-	-	12/12	-	-	COSM1180918,TMP_ESP_20_31022442_31022442
26	INDEL	chr20	31022441	A	+G	198	127	71	0.353	ASXL1	ENSG00000171456	FRAME_SHIFT	-	-	13/13	YES	-	COSM1180918,TMP_ESP_20_31022442_31022442
26	INDEL	chr20	31022441	A	+G	198	127	71	0.353	ASXL1	ENSG00000171456	FRAME_SHIFT	-	-	12/12	-	-	COSM1180918,TMP_ESP_20_31022442_31022442
29	SNV	chr20	31022847	C	T	516	308	208	0.403	ASXL1	ENSG00000171456	STOP_GAINED	ENST00000375687.4:c.2332C>T	ENSP00000364839.4:p.Gln778Ter	13/13	YES	-	COSM133576
29	SNV	chr20	31022847	C	T	516	308	208	0.403	ASXL1	ENSG00000171456	STOP_GAINED	ENST00000306058.5:c.2317C>T	ENSP00000305119.5:p.Gln773Ter	12/12	-	-	COSM133576
30	INDEL	chr20	31022441	A	+G	144	101	43	0.297	ASXL1	ENSG00000171456	FRAME_SHIFT	-	-	12/12	-	-	COSM1180918,TMP_ESP_20_31022442_31022442
30	INDEL	chr20	31022441	A	+G	144	101	43	0.297	ASXL1	ENSG00000171456	FRAME_SHIFT	-	-	13/13	YES	-	COSM1180918,TMP_ESP_20_31022442_31022442
34	INDEL	chr20	31022441	A	+G	192	140	52	0.264	ASXL1	ENSG00000171456	FRAME_SHIFT	-	-	13/13	YES	-	COSM1180918,TMP_ESP_20_31022442_31022442
34	INDEL	chr20	31022441	A	+G	192	140	52	0.264	ASXL1	ENSG00000171456	FRAME_SHIFT	-	-	12/12	-	-	COSM1180918,TMP_ESP_20_31022442_31022442
36	INDEL	chr20	31022441	A	+G	266	152	114	0.422	ASXL1	ENSG00000171456	FRAME_SHIFT	-	-	12/12	-	-	COSM1180918,TMP_ESP_20_31022442_31022442
36	SNV	chr20	31022947	A	G	557	297	260	0.467	ASXL1	ENSG00000171456	NON_SYNONYMOUS_CODING	ENST00000375687.4:c.2432A>G	ENSP00000364839.4:p.Asn811Ser	13/13	YES	-	COSM1411085
36	INDEL	chr20	31022441	A	+G	266	152	114	0.422	ASXL1	ENSG00000171456	FRAME_SHIFT	-	-	13/13	YES	-	COSM1180918,TMP_ESP_20_31022442_31022442
36	SNV	chr20	31022947	A	G	557	297	260	0.467	ASXL1	ENSG00000171456	NON_SYNONYMOUS_CODING	ENST00000306058.5:c.2417A>G	ENSP00000305119.5:p.Asn806Ser	12/12	-	-	COSM1411085
44	SNV	chr20	31023408	C	T	651	410	241	0.37	ASXL1	ENSG00000171456	STOP_GAINED	ENST00000375687.4:c.2893C>T	ENSP00000364839.4:p.Arg965Ter	13/13	YES	-	rs397515401,COSM267971
44	SNV	chr20	31023408	C	T	651	410	241	0.37	ASXL1	ENSG00000171456	STOP_GAINED	ENST00000306058.5:c.2878C>T	ENSP00000305119.5:p.Arg960Ter	12/12	-	-	rs397515401,COSM267971
S03	INDEL	chr20	31021157	T	-TG	713	372	341	0.478	ASXL1	ENSG00000171456	FRAME_SHIFT	ENST00000306058.5:c.1142delTG	ENSP00000305119.5:p.Leu381CysfsTer76	11/12	-	-	-
S03	INDEL	chr20	31021157	T	-TG	713	372	341	0.478	ASXL1	ENSG00000171456	FRAME_SHIFT	ENST00000553345.1:c.473delTG	ENSP00000451444.1:p.Leu158CysfsTer?	5/5	-	-	-
S03	INDEL	chr20	31021157	T	-TG	713	372	341	0.478	ASXL1	ENSG00000171456	FRAME_SHIFT	ENST00000375687.4:c.1157delTG	ENSP00000364839.4:p.Leu386CysfsTer76	12/13	YES	-	-
S10	SNV	chr20	31024458	C	T	596	307	289	0.485	ASXL1	ENSG00000171456	STOP_GAINED	ENST00000306058.5:c.3928C>T	ENSP00000305119.5:p.Gln1310Ter	12/12	-	-	-
S10	SNV	chr20	31024458	C	T	596	307	289	0.485	ASXL1	ENSG00000171456	STOP_GAINED	ENST00000375687.4:c.3943C>T	ENSP00000364839.4:p.Gln1315Ter	13/13	YES	-	-
S12	INDEL	chr20	31022402	T	- CACCACTGCCATAGA GAGGCGGC	280	199	81	0.289	ASXL1	ENSG00000171456	FRAME_SHIFT	ENST00000306058.5:c.1873delC	ENSP00000305119.5:p.His625ThrfsTer73	12/12	-	-	-
S12	INDEL	chr20	31022402	T	- CACCACTGCCATAGA GAGGCGGC	280	199	81	0.289	ASXL1	ENSG00000171456	FRAME_SHIFT	ENST00000375687.4:c.1888delC	ENSP00000364839.4:p.His630ThrfsTer73	13/13	YES	-	-
S17	SNV	chr20	31022288	C	A	730	409	321	0.44	ASXL1	ENSG00000171456	STOP_GAINED	ENST00000375687.4:c.1773C>A	ENSP00000364839.4:p.Tyr591Ter	13/13	YES	-	rs371369583,COSM1681609,COSM53200
S17	SNV	chr20	31022288	C	A	730	409	321	0.44	ASXL1	ENSG00000171456	STOP_GAINED	ENST00000306058.5:c.1758C>A	ENSP00000305119.5:p.Tyr586Ter	12/12	-	-	rs371369583,COSM1681609,COSM53200
S36	SNV	chr20	31022827	T	A	885	475	410	0.463	ASXL1	ENSG00000171456	NON_SYNONYMOUS_CODING	ENST00000306058.5:c.2297T>A	ENSP00000305119.5:p.Val766Glu	12/12	-	-	-
S36	INDEL	chr20	31022825	A	-G	897	482	415	0.463	ASXL1	ENSG00000171456	FRAME_SHIFT	ENST00000306058.5:c.2296delG	ENSP00000305119.5:p.Val766Ter	12/12	-	-	-
S36	SNV	chr20	31022827	T	A	885	475	410	0.463	ASXL1	ENSG00000171456	NON_SYNONYMOUS_CODING	ENST00000375687.4:c.2312T>A	ENSP00000364839.4:p.Val771Glu	13/13	YES	-	-
S36	INDEL	chr20	31022825	A	-G	897	482	415	0.463	ASXL1	ENSG00000171456	FRAME_SHIFT	ENST00000375687.4:c.2311delG	ENSP00000364839.4:p.Val771Ter	13/13	YES	-	-
S37	SNV	chr20	31022581	C	G	628	413	215	0.342	ASXL1	ENSG00000171456	STOP_GAINED	ENST00000306058.5:c.2051C>G	ENSP00000305119.5:p.Ser684Ter	12/12	-	-	COSM133037
S37	SNV	chr20	31022581	C	G	628	413	215	0.342	ASXL1	ENSG00000171456	STOP_GAINED	ENST00000375687.4:c.2066C>G	ENSP00000364839.4:p.Ser689Ter	13/13	YES	-	COSM133037
S51	INDEL	chr20	31022440	G	+A	352	195	157	0.446	ASXL1	ENSG00000171456	FRAME_SHIFT	-	-	12/12	-	-	COSM3737035
S51	INDEL	chr20	31022440	G	+A	352	195	157	0.446	ASXL1	ENSG00000171456	FRAME_SHIFT	-	-	13/13	YES	-	COSM3737035
S56	INDEL	chr20	31022441	A	+G	541	338	203	0.375	ASXL1	ENSG00000171456	FRAME_SHIFT	-	-	13/13	YES	-	COSM1180918,TMP_ESP_20_31022442_31022442
S56	INDEL	chr20	31022441	A	+G	541	338	203	0.375	ASXL1	ENSG00000171456	FRAME_SHIFT	-	-	12/12	-	-	COSM1180918,TMP_ESP_20_31022442_31022442
S58	INDEL	chr20	31022441	A	+G	453	255	198	0.431	ASXL1	ENSG00000171456	FRAME_SHIFT	-	-	13/13	YES	-	COSM1180918,TMP_ESP_20_31022442_31022442
S58	INDEL	chr20	31022441	A	+G	453	255	198	0.431	ASXL1	ENSG00000171456	FRAME_SHIFT	-	-	12/12	-	-	COSM1180918,TMP_ESP_20_31022442_31022442
S61	INDEL	chr20	31022441	A	+G	431	240	191	0.443	ASXL1	ENSG00000171456	FRAME_SHIFT	-	-	13/13	YES	-	COSM1180918,TMP_ESP_20_31022442_31022442
S61	INDEL	chr20	31022441	A	+G	431	240	191	0.443	ASXL1	ENSG00000171456	FRAME_SHIFT	-	-	12/12	-	-	COSM1180918,TMP_ESP_20_31022442_31022442
S64	INDEL	chr20	31022441	A	+G	334	200	134	0.4	ASXL1	ENSG00000171456	FRAME_SHIFT	-	-	13/13	YES	-	COSM1180918,TMP_ESP_20_31022442_31022442
S64	INDEL	chr20	31022441	A	+G	334	200	134	0.4	ASXL1	ENSG00000171456	FRAME_SHIFT	-	-	12/12	-	-	COSM1180918,TMP_ESP_20_31022442_31022442
S22	SNV	chr2	216213859	C	T	552	296	256	0.464	ATIC	ENSG00000138363	NON_SYNONYMOUS_CODING	ENST00000540518.1:c.1369C>T	ENSP00000440523.1:p.Pro457Ser	15/16	-	-	COSM3577496
S22	SNV	chr2	216213859	C	T	552	296	256	0.464	ATIC	ENSG00000138363	NON_SYNONYMOUS_CODING	ENST00000435675.1:c.1543C>T	ENSP00000415935.1:p.Pro515Ser	14/15	-	-	COSM3577496
S22	SNV	chr2	216213859	C	T	552	296	256	0.464	ATIC	ENSG00000138363	NON_SYNONYMOUS_CODING	ENST00000236959.9:c.1546C>T	ENSP00000236959.9:p.Pro516Ser	15/16	YES	-	COSM3577496
S22	SNV	chr2	216213859	C	T	552	296	256	0.464	ATIC	ENSG00000138363	NON_SYNONYMOUS_CODING	ENST00000442048.1:c.91C>T	ENSP00000391399.1:p.Pro31Ser	2/3	-	-	COSM3577496
S22	SNV	chr2	216213859	C	T	552	296	256	0.464	ATIC	ENSG00000138363	NON_SYNONYMOUS_CODING	ENST00000426233.1:c.551C>T	ENSP00000401936.1:p.Pro185Ser	5/5	-	-	COSM3577496
S22	SNV	chr2	216213414	G	A	149	71	78	0.524	ATIC	ENSG00000138363	NON_SYNONYMOUS_CODING	ENST00000442048.1:c.8G>A	ENSP00000391399.1:p.Arg3Gln	1/3	-	0.0004	rs560702336
32	SNV	chr11	108183218	G	A	62	39	23	0.371	ATM	ENSG00000149311	NON_SYNONYMOUS_CODING	ENST00000452508.2:c.5999G>A	ENSP00000388058.2:p.Ser2000Asn	41/64	-	-	-
32	SNV	chr11	108183218	G	A	62	39	23	0.371	ATM	ENSG00000149311	NON_SYNONYMOUS_CODING	ENST00000278616.4:c.5999G>A	ENSP00000278616.4:p.Ser2000Asn	40/63	YES	-	-
48	SNV	chr11	108214073	C	A	255	136	119	0.467	ATM	ENSG00000149311	NON_SYNONYMOUS_CODING	ENST00000278616.4:c.8393C>A	ENSP00000278616.4:p.Ala2798Asp	57/63	YES	-	-
48	SNV	chr11	108214073	C	A	255	136	119	0.467	ATM	ENSG00000149311	NON_SYNONYMOUS_CODING	ENST00000452508.2:c.8393C>A	ENSP00000388058.2:p.Ala2798Asp	58/64	-	-	-
10	SNV	chr7	70255459	G	A	233	152	81	0.348	AUTS2	ENSG00000158321	NON_SYNONYMOUS_CODING	ENST00000406775.2:c.3185G>A	ENSP00000385263.2:p.Arg1062Gln	18/18	-	-	-
10	SNV	chr7	70255459	G	A	233	152	81	0.348	AUTS2	ENSG00000158321	NON_SYNONYMOUS_CODING	ENST00000342771.4:c.3257G>A	ENSP00000344087.4:p.Arg1086Gln	19/19	YES	-	-
19	SNV	chr7	70242110	A	G	818	455	363	0.444	AUTS2	ENSG00000158321	NON_SYNONYMOUS_CODING	ENST00000406775.2:c.1882A>G	ENSP00000385263.2:p.Met628Val	13/18	-	-	-
19	SNV	chr7	70242110	A	G	818	455	363	0.444	AUTS2	ENSG00000158321	NON_SYNONYMOUS_CODING	ENST00000342771.4:c.1954A>G	ENSP00000344087.4:p.Met652Val	14/19	YES	-	-
S25	SNV	chr7	70250956	C	T	90	61	29	0.322	AUTS2	ENSG00000158321	NON_SYNONYMOUS_CODING	ENST00000418686.1:c.80C>T	ENSP00000392333.1:p.Pro27Leu	2/4	-	-	-
S25	SNV	chr7	70250956	C	T	90	61	29	0.322	AUTS2	ENSG00000158321	NON_SYNONYMOUS_CODING	ENST00000342771.4:c.2240C>T	ENSP00000344087.4:p.Pro747Leu	17/19	YES	-	-
S25	SNV	chr7	70250956	C	T	90	61	29	0.322	AUTS2	ENSG00000158321	NON_SYNONYMOUS_CODING	ENST00000406775.2:c.2168C>T	ENSP00000385263.2:p.Pro723Leu	16/18	-	-	-
S42	SNV	chr7	70246662	G	A	511	275	236	0.462	AUTS2	ENSG00000158321	NON_SYNONYMOUS_CODING	ENST00000406775.2:c.1994G>A	ENSP00000385263.2:p.Arg665His	14/18	-	-	-
S42	SNV	chr7	70246662	G	A	511	275	236	0.462	AUTS2	ENSG00000158321	NON_SYNONYMOUS_CODING	ENST00000342771.4:c.2066G>A	ENSP00000344087.4:p.Arg689His	15/19	YES	-	-
28	SNV	chr3	52437803	T	C	183	92	91	0.497	BAP1	ENSG00000163930	NON_SYNONYMOUS_CODING	ENST00000296288.5:c.1304A>G	ENSP00000296288.5:p.Lys435Arg	13/17	-	-	-
28	SNV	chr3	52437803	T	C	183	92	91	0.497	BAP1	ENSG00000163930	NON_SYNONYMOUS_CODING	ENST00000460680.1:c.1358A>G	ENSP00000417132.1:p.Lys453Arg	13/17	YES	-	-
S62	SNV	chr1	85733617	T	C	675	364	311	0.461	BCL10	ENSG00000142867	NON_SYNONYMOUS_CODING	ENST00000370580.1:c.395A>G	ENSP00000359612.1:p.Asn132Ser	3/3	YES	-	-
25	SNV	chr2	60687781	T	G	340	210	130	0.382	BCL11A	ENSG00000119866	NON_SYNONYMOUS_CODING	ENST00000358510.4:c.2164A>C	ENSP00000351307.4:p.Asn722His	3/3	-	-	-
25	SNV	chr2	60687781	T	G	340	210	130	0.382	BCL11A	ENSG00000119866	NON_SYNONYMOUS_CODING	ENST00000538214.1:c.2164A>C	ENSP00000438303.1:p.Asn722His	3/4	-	-	-
25	SNV	chr2	60687781	T	G	340	210	130	0.382	BCL11A	ENSG00000119866	NON_SYNONYMOUS_CODING	ENST00000335712.6:c.2266A>C	ENSP00000338774.6:p.Asn756His	4/4	YES	-	-
46	SNV	chr2	60687652	C	T	170	93	77	0.453	BCL11A	ENSG00000119866	NON_SYNONYMOUS_CODING	ENST00000335712.6:c.2395G>A	ENSP00000338774.6:p.Val799Ile	4/4	YES	-	-
46	SNV	chr2	60687652	C	T	170	93	77	0.453	BCL11A	ENSG00000119866	NON_SYNONYMOUS_CODING	ENST00000358510.4:c.2293G>A	ENSP00000351307.4:p.Val765Ile	3/3	-	-	-
1	INDEL	chr14	99737635	G	+CGCGCTGC	59	41	18	0.3	BCL11B	ENSG00000127152	UTR_5_PRIME	-	-	1/2	-	-	-
1	INDEL	chr14	99737635	G	+CGCGCTGC	59	41	18	0.3	BCL11B	ENSG00000127152	UTR_5_PRIME	-	-	1/3	-	-	-
S60	SNV	chr14	99642124	C	T	358	200	158	0.441	BCL11B	ENSG00000127152	NON_SYNONYMOUS_CODING	ENST00000443726.2:c.467G>A	ENSP00000387419.2:p.Arg156His	2/2	-	-	-
S60	SNV	chr14	99642124	C	T	358	200	158	0.441	BCL11B	ENSG00000127152	NON_SYNONYMOUS_CODING	ENST00000345514.2:c.836G>A	ENSP00000280435.6:p.Arg279His	3/3	-	-	-
S60	SNV	chr14	99642124	C	T	358	200	158	0.441	BCL11B	ENSG00000127152	NON_SYNONYMOUS_CODING	ENST00000357195.3:c.1049G>A	ENSP00000349723.3:p.Arg350His	4/4	YES	-	-
33	SNV	chr1	147088390	G	A	307	160	147	0.479	BCL9	ENSG00000116128	NON_SYNONYMOUS_CODING	ENST00000234739.3:c.535G>A	ENSP00000234739.3:p.Val179Met	6/10	YES	-	-
21	INDEL	chrX	39933791	G	+T	432	260	172	0.397	BCOR	ENSG00000183337	FRAME_SHIFT	-	-	4/9	-	-	-

21	INDEL	chrX	39933791	G	+T	432	260	172	0.397	BCOR	ENSG00000183337	FRAME_SHIFT	-	-	4/15	YES	-	-	
21	INDEL	chrX	39933791	G	+T	432	260	172	0.397	BCOR	ENSG00000183337	FRAME_SHIFT	-	-	4/14	-	-	-	
21	INDEL	chrX	39933791	G	+T	432	260	172	0.397	BCOR	ENSG00000183337	FRAME_SHIFT	-	-	4/15	-	-	-	
21	INDEL	chrX	39933791	G	+T	432	260	172	0.397	BCOR	ENSG00000183337	FRAME_SHIFT	-	-	4/15	-	-	-	
30	INDEL	chrX	39913178	A	+G	256	136	120	0.469	BCOR	ENSG00000183337	FRAME_SHIFT	-	-	6/7	-	-	-	
30	INDEL	chrX	39913178	A	+G	256	136	120	0.469	BCOR	ENSG00000183337	FRAME_SHIFT	-	-	13/14	-	-	-	
30	INDEL	chrX	39913178	A	+G	256	136	120	0.469	BCOR	ENSG00000183337	FRAME_SHIFT	-	-	14/15	YES	-	-	
30	INDEL	chrX	39913178	A	+G	256	136	120	0.469	BCOR	ENSG00000183337	FRAME_SHIFT	-	-	14/15	-	-	-	
30	INDEL	chrX	39913178	A	+G	256	136	120	0.469	BCOR	ENSG00000183337	FRAME_SHIFT	-	-	14/15	-	-	-	
30	INDEL	chrX	39913178	A	+G	256	136	120	0.469	BCOR	ENSG00000183337	FRAME_SHIFT	-	-	9/11	-	-	-	
30	INDEL	chrX	39913178	A	+G	256	136	120	0.469	BCOR	ENSG00000183337	FRAME_SHIFT	-	-	8/10	-	-	-	
34	INDEL	chrX	39934162	C	-TGAA	366	279	87	0.234	BCOR	ENSG00000183337	FRAME_SHIFT	ENST00000378455.4:c.437delG	ENSP00000367716.4:p.Ser146MetfsTer15	4/14	-	-	-	
34	INDEL	chrX	39934156	A	-TAG	393	306	87	0.221	BCOR	ENSG00000183337	CODON_CHANGE_PLUS_CODON_DELETION	ENST00000397354.3:c.443delT	ENSP00000380512.3:p.Ile148AsnfsTer13	4/15	-	-	-	
34	INDEL	chrX	39934160	C	-A	373	286	87	0.233	BCOR	ENSG00000183337	FRAME_SHIFT	ENST00000378455.4:c.439delG	ENSP00000367716.4:p.Ala147LeufsTer14	4/14	-	-	-	
34	INDEL	chrX	39934160	C	-A	373	286	87	0.233	BCOR	ENSG00000183337	FRAME_SHIFT	ENST00000342274.4:c.439delG	ENSP00000345923.4:p.Ala147LeufsTer14	4/15	-	-	-	
34	INDEL	chrX	39934160	C	-A	373	286	87	0.233	BCOR	ENSG00000183337	FRAME_SHIFT	ENST00000406200.2:c.439delG	ENSP00000384485.2:p.Ala147LeufsTer14	4/9	-	-	-	
34	INDEL	chrX	39934156	A	-TAG	393	306	87	0.221	BCOR	ENSG00000183337	CODON_CHANGE_PLUS_CODON_DELETION	ENST00000378444.4:c.443delT	ENSP00000367705.4:p.Ile148AsnfsTer13	4/15	YES	-	-	
34	INDEL	chrX	39934160	C	-A	373	286	87	0.233	BCOR	ENSG00000183337	FRAME_SHIFT	ENST00000397354.3:c.439delG	ENSP00000380512.3:p.Ala147LeufsTer14	4/15	-	-	-	
34	INDEL	chrX	39934162	C	-TGAA	366	279	87	0.234	BCOR	ENSG00000183337	FRAME_SHIFT	ENST00000378444.4:c.437delG	ENSP00000367705.4:p.Ser146MetfsTer15	4/15	YES	-	-	
34	INDEL	chrX	39934156	A	-TAG	393	306	87	0.221	BCOR	ENSG00000183337	CODON_CHANGE_PLUS_CODON_DELETION	ENST00000406200.2:c.443delT	ENSP00000384485.2:p.Ile148AsnfsTer13	4/9	-	-	-	
34	INDEL	chrX	39934156	A	-TAG	393	306	87	0.221	BCOR	ENSG00000183337	CODON_CHANGE_PLUS_CODON_DELETION	ENST00000378455.4:c.443delT	ENSP00000367716.4:p.Ile148AsnfsTer13	4/14	-	-	-	
34	INDEL	chrX	39934162	C	-TGAA	366	279	87	0.234	BCOR	ENSG00000183337	FRAME_SHIFT	ENST00000397354.3:c.437delG	ENSP00000380512.3:p.Ser146MetfsTer15	4/15	-	-	-	
34	INDEL	chrX	39934160	C	-A	373	286	87	0.233	BCOR	ENSG00000183337	FRAME_SHIFT	ENST00000378444.4:c.439delG	ENSP00000367705.4:p.Ala147LeufsTer14	4/15	YES	-	-	
34	INDEL	chrX	39934156	A	-TAG	393	306	87	0.221	BCOR	ENSG00000183337	CODON_CHANGE_PLUS_CODON_DELETION	ENST00000342274.4:c.443delT	ENSP00000345923.4:p.Ile148AsnfsTer13	4/15	-	-	-	
34	INDEL	chrX	39934162	C	-TGAA	366	279	87	0.234	BCOR	ENSG00000183337	FRAME_SHIFT	ENST00000342274.4:c.437delG	ENSP00000345923.4:p.Ser146MetfsTer15	4/15	-	-	-	
34	INDEL	chrX	39934162	C	-TGAA	366	279	87	0.234	BCOR	ENSG00000183337	FRAME_SHIFT	ENST00000406200.2:c.437delG	ENSP00000384485.2:p.Ser146MetfsTer15	4/9	-	-	-	
44	SNV	chrX	39931673	G	A	358	263	95	0.265	BCOR	ENSG00000183337	STOP_GAINED	ENST00000378455.4:c.2926C>T	ENSP00000367716.4:p.Arg976Ter	4/14	-	-	rs121434619,CM040984	
44	SNV	chrX	39931673	G	A	358	263	95	0.265	BCOR	ENSG00000183337	STOP_GAINED	ENST00000378444.4:c.2926C>T	ENSP00000367705.4:p.Arg976Ter	4/15	YES	-	rs121434619,CM040984	
44	SNV	chrX	39931673	G	A	358	263	95	0.265	BCOR	ENSG00000183337	STOP_GAINED	ENST00000342274.4:c.2926C>T	ENSP00000345923.4:p.Arg976Ter	4/15	-	-	rs121434619,CM040984	
44	SNV	chrX	39931673	G	A	358	263	95	0.265	BCOR	ENSG00000183337	STOP_GAINED	ENST00000406200.2:c.2926C>T	ENSP00000384485.2:p.Arg976Ter	4/9	-	-	rs121434619,CM040984	
44	SNV	chrX	39931673	G	A	358	263	95	0.265	BCOR	ENSG00000183337	STOP_GAINED	ENST00000397354.3:c.2926C>T	ENSP00000380512.3:p.Arg976Ter	4/15	-	-	rs121434619,CM040984	
47	INDEL	chrX	39932628	G	+T	670	360	310	0.463	BCOR	ENSG00000183337	FRAME_SHIFT	-	-	4/9	-	-	-	
47	INDEL	chrX	39932628	G	+T	670	360	310	0.463	BCOR	ENSG00000183337	FRAME_SHIFT	-	-	4/15	YES	-	-	
47	INDEL	chrX	39932628	G	+T	670	360	310	0.463	BCOR	ENSG00000183337	FRAME_SHIFT	-	-	4/14	-	-	-	
47	INDEL	chrX	39932628	G	+T	670	360	310	0.463	BCOR	ENSG00000183337	FRAME_SHIFT	-	-	4/15	-	-	-	
47	INDEL	chrX	39932628	G	+T	670	360	310	0.463	BCOR	ENSG00000183337	FRAME_SHIFT	-	-	4/15	-	-	-	
S03	INDEL	chrX	39913140	C	-CT	240	19	221	0.921	BCOR	ENSG00000183337	SPLICE_SITE_REGION	ENST00000378444.4:c.4976+1delG	-	14/15	YES	-	-	
S03	INDEL	chrX	39913140	C	-CT	240	19	221	0.921	BCOR	ENSG00000183337	FRAME_SHIFT	ENST00000378455.4:c.4820+1delG	-	13/14	-	-	-	
S03	INDEL	chrX	39913140	C	-CT	240	19	221	0.921	BCOR	ENSG00000183337	FRAME_SHIFT	ENST00000378463.1:c.1505+1delG	-	9/11	-	-	-	
S03	INDEL	chrX	39913140	C	-CT	240	19	221	0.921	BCOR	ENSG00000183337	FRAME_SHIFT	ENST00000342274.4:c.4874+1delG	-	14/15	-	-	-	
S03	INDEL	chrX	39913140	C	-CT	240	19	221	0.921	BCOR	ENSG00000183337	SPLICE_SITE_REGION	ENST00000413905.1:c.1586+1delG	-	8/10	-	-	-	
S03	INDEL	chrX	39913140	C	-CT	240	19	221	0.921	BCOR	ENSG00000183337	FRAME_SHIFT	ENST00000397354.3:c.4874+1delG	-	14/15	-	-	-	
S03	INDEL	chrX	39913140	C	-CT	240	19	221	0.921	BCOR	ENSG00000183337	FRAME_SHIFT	ENST00000378444.4:c.4976+1delG	-	14/15	YES	-	-	
S03	INDEL	chrX	39913140	C	-CT	240	19	221	0.921	BCOR	ENSG00000183337	FRAME_SHIFT	ENST00000413905.1:c.1586+1delG	-	8/10	-	-	-	
S03	INDEL	chrX	39913140	C	-CT	240	19	221	0.921	BCOR	ENSG00000183337	SPLICE_SITE_REGION	ENST00000397354.3:c.4874+1delG	-	14/15	-	-	-	
S03	INDEL	chrX	39913140	C	-CT	240	19	221	0.921	BCOR	ENSG00000183337	FRAME_SHIFT	ENST00000442018.1:c.996delG	ENSP00000387552.1:p.Ter333GlufsTer10	6/7	-	-	-	
S03	INDEL	chrX	39913140	C	-CT	240	19	221	0.921	BCOR	ENSG00000183337	SPLICE_SITE_REGION	ENST00000378455.4:c.4820+1delG	-	13/14	-	-	-	
S03	INDEL	chrX	39913140	C	-CT	240	19	221	0.921	BCOR	ENSG00000183337	SPLICE_SITE_REGION	ENST00000378463.1:c.1505+1delG	-	9/11	-	-	-	
S03	INDEL	chrX	39913140	C	-CT	240	19	221	0.921	BCOR	ENSG00000183337	SPLICE_SITE_REGION	ENST00000342274.4:c.4874+1delG	-	14/15	-	-	-	
S27	SNV	chrX	39923050	G	A	895	615	280	0.313	BCOR	ENSG00000183337	STOP_GAINED	ENST00000378455.4:c.3502C>T	ENSP00000367716.4:p.Gln1168Ter	7/14	-	-	-	
S27	SNV	chrX	39923050	G	A	895	615	280	0.313	BCOR	ENSG00000183337	STOP_GAINED	ENST00000342274.4:c.3556C>T	ENSP00000345923.4:p.Gln1186Ter	8/15	-	-	-	
S27	SNV	chrX	39923050	G	A	895	615	280	0.313	BCOR	ENSG00000183337	STOP_GAINED	ENST00000397354.3:c.3556C>T	ENSP00000380512.3:p.Gln1186Ter	8/15	-	-	-	
S27	SNV	chrX	39923050	G	A	895	615	280	0.313	BCOR	ENSG00000183337	STOP_GAINED	ENST00000378463.1:c.187C>T	ENSP00000367724.1:p.Gln63Ter	3/11	-	-	-	
S27	SNV	chrX	39923050	G	A	895	615	280	0.313	BCOR	ENSG00000183337	STOP_GAINED	ENST00000406200.2:c.3556C>T	ENSP00000384485.2:p.Gln1186Ter	8/9	-	-	-	
S27	SNV	chrX	39923050	G	A	895	615	280	0.313	BCOR	ENSG00000183337	STOP_GAINED	ENST00000378444.4:c.3658C>T	ENSP00000367705.4:p.Gln1220Ter	8/15	YES	-	-	
S27	SNV	chrX	39923050	G	A	895	615	280	0.313	BCOR	ENSG00000183337	STOP_GAINED	ENST00000413905.1:c.268C>T	ENSP00000408006.1:p.Gln90Ter	2/10	-	-	-	
S29	SNV	chrX	39923601	G	A	110	14	96	0.873	BCOR	ENSG00000183337	STOP_GAINED	ENST00000378463.1:c.121C>T	ENSP00000367724.1:p.Arg41Ter	2/11	-	-	-	
S29	SNV	chrX	39923601	G	A	110	14	96	0.873	BCOR	ENSG00000183337	STOP_GAINED	ENST00000342274.4:c.3490C>T	ENSP00000345923.4:p.Arg1164Ter	7/15	-	-	-	
S29	SNV	chrX	39923601	G	A	110	14	96	0.873	BCOR	ENSG00000183337	STOP_GAINED	ENST00000378444.4:c.3490C>T	ENSP00000367705.4:p.Arg1164Ter	7/15	YES	-	-	
S29	SNV	chrX	39923601	G	A	110	14	96	0.873	BCOR	ENSG00000183337	STOP_GAINED	ENST00000378455.4:c.3436C>T	ENSP00000367716.4:p.Arg1146Ter	6/14	-	-	-	
S29	SNV	chrX	39923601	G	A	110	14	96	0.873	BCOR	ENSG00000183337	STOP_GAINED	ENST00000397354.3:c.3490C>T	ENSP00000380512.3:p.Arg1164Ter	7/15	-	-	-	
S29	SNV	chrX	39923601	G	A	110	14	96	0.873	BCOR	ENSG00000183337	STOP_GAINED	ENST00000413905.1:c.202C>T	ENSP00000408006.1:p.Arg68Ter	1/10	-	-	-	
S29	SNV	chrX	39923601	G	A	110	14	96	0.873	BCOR	ENSG00000183337	STOP_GAINED	ENST00000406200.2:c.3490C>T	ENSP00000384485.2:p.Arg1164Ter	7/9	-	-	-	
S51	SNV	chrX	39932312	G	A	423	1	422	0.998	BCOR	ENSG00000183337	NON_SYNONYMOUS_CODING	ENST00000378455.4:c.2287C>T	ENSP00000367716.4:p.Arg763Trp	4/14	-	-	-	
S51	SNV	chrX	39932312	G	A	423	1	422	0.998	BCOR	ENSG00000183337	NON_SYNONYMOUS_CODING	ENST00000342274.4:c.2287C>T	ENSP00000345923.4:p.Arg763Trp	4/15	-	-	-	
S51	SNV	chrX	39932312	G	A	423	1	422	0.998	BCOR	ENSG00000183337	NON_SYNONYMOUS_CODING	ENST00000378444.4:c.2287C>T	ENSP00000367705.4:p.Arg763Trp	4/15	YES	-	-	
S51	SNV	chrX	39932312	G	A	423	1	422	0.998	BCOR	ENSG00000183337	NON_SYNONYMOUS_CODING	ENST00000406200.2:c.2287C>T	ENSP00000384485.2:p.Arg763Trp	4/9	-	-	-	
S51	SNV	chrX	39932312	G	A	423	1	422	0.998	BCOR	ENSG00000183337	NON_SYNONYMOUS_CODING	ENST00000397354.3:c.2287C>T	ENSP00000380512.3:p.Arg763Trp	4/15	-	-	-	
55	SNV	chrX	129185888	G	A	246	2	244	0.988	BCORL1	ENSG00000085185	NON_SYNONYMOUS_CODING	ENST00000340052.1:c.4750G>A	ENSP00000437778.2:p.Asp1584Asn	11/12	YES	-	COSM1682882	
55	SNV	chrX	129185888	G	A	246	2	244	0.988	BCORL1	ENSG00000085185	NON_SYNONYMOUS_CODING	ENST00000218147.7:c.4750G>A	ENSP00000218147.7:p.Asp1584Asn	12/13	-	-	COSM1682882	
55	SNV	chrX	129185888	G	A	246	2	244	0.988	BCORL1	ENSG00000085185	NON_SYNONYMOUS_CODING	ENST00000330743.5:c.4972G>A	ENSP00000307541.5:p.Asp1658Asn	13/14	-	-	COSM1682882	
55	SNV	chrX	129185888	G	A	246	2	244	0.988	BCORL1	ENSG00000085185	NON_SYNONYMOUS_CODING	ENST00000456822.1:c.3772G>A	ENSP00000399483.1:p.Asp1258Asn	10/11	-	-	COSM1682882	
55	SNV	chrX	129185888	G	A	246	2	244	0.988	BCORL1	ENSG00000085185	NON_SYNONYMOUS_CODING	ENST00000359304.2:c.4360G>A	ENSP00000352253.2:p.Asp1454Asn	11/12	-	-	COSM1682882	
S55	SNV	chr22	23603177	G	A	990	708	282	0.284	BCR	ENSG00000186716	STOP_GAINED	ENST00000359540.3:c.1502G>A	ENSP00000352535.3:p.Trp501Ter	3/22	-	-	-	
S55	SNV	chr22	23603177	G	A	990	708	282	0.284	BCR	ENSG00000186716	STOP_GAINED	ENST00000427791.1:c.62G>A	ENSP00000396531.1:p.Trp21Ter	2/8	-	-	-	
S55	SNV	chr22	23603177	G	A	990	708	282	0.284	BCR	ENSG00000186716	STOP_GAINED	ENST00000305877.8:c.1502G>A	ENSP00000303507.8:p.Trp501Ter	3/23	YES	-	-	
38	SNV	chr17	40962764	C	G	453	227	226	0.499	BECN1	ENSG00000126581	UTR_3_PRIME	ENST00000438274.3:c.*156G>C	-	9/9	-	-	-	
38	SNV	chr17	40962764	C	G	453	227	226	0.499	BECN1	ENSG00000126581	UTR_3_PRIME	ENST00000361523.4:c.*14G>C	-	12/12	YES	-	-	
S61	SNV	chr11	102201893	T	A	565	275	290	0.513	BIRC3	ENSG00000023445	NON_SYNONYMOUS_CODING	ENST00000263464.3:c.1245T>A	ENSP00000263464.3:p.Asp415Glu	6/9	YES	-	-	
S61	SNV	chr11	102201893	T	A	565	275	290	0.513	BIRC3	ENSG00000023445	NON_SYNONYMOUS_CODING	ENST00000532808.1:c.1245T>A	ENSP00000432907.1:p.Asp415Glu	7/10	-	-	-	
S40	SNV	chr15	91303500	G	A	474	246	228	0.481	BLM	ENSG00000197299	NON_SYNONYMOUS_CODING	ENST00000355112.3:c.1211G>A	ENSP00000347232.3:p.Arg404Gln	6/22	YES	-	-	
S40	SNV	chr15	91303500	G	A	474	246	228	0.481	BLM	ENSG00000197299	NON_SYNONYMOUS_CODING	ENST00000560509.1:c.1211G>A	ENSP00000454158.1:p.Arg404Gln	6/20	-	-	-	
1	SNV	chr7	140624440	C	T	30	15	15	0.5	BRAF	ENSG00000157764	NON_SYNONYMOUS_CODING	ENST00000288602.6:c.64G>A	ENSP00000288602.6:p.Asp22Asn	1/18	YES	-	rs397507456	
7	SNV	chr7	140508726	G	C	573	321	252	0.44	BRAF	ENSG00000157764	NON_SYNONYMOUS_CODING	ENST00000288602.6:c.574C>G	ENSP00000288602.6:p.Pro192Ala	4/18	YES	-	-	
26	SNV	chr13	32913726	T	A	283	161	122	0.431	BRCA2	ENSG00000139618	NON_SYNONYMOUS_CODING	ENST00000544455.1:c.5234T>A	ENSP00000439902.1:p.Met1745Lys	11/28	YES	-	-	
26	SNV	chr13	32913726	T	A	283	161	122	0.431	BRCA2	ENSG00000139618	NON_SYNONYMOUS_CODING	ENST00000380152.3:c.5234T>A	ENSP00000369497.3:p.Met1745Lys	11/27	-	-	-	
43	SNV	chr13	32907520	T	G	215	118	97	0.451	BRCA2	ENSG00000139618	NON_SYNONYMOUS_CODING	ENST00000380152.3:c.1905T>G	ENSP00000369497.3:p.Asp635Glu	10/27	-	-	-	
43	SNV	chr13	32907520	T	G	215	118	97	0.451	BRCA2	ENSG00000139618	NON_SYNONYMOUS_CODING	ENST00000544455.1:c.1905T>G	ENSP00000439902.1:p.Asp635Glu	10/28	YES	-	-	
S37	SNV	chr13	32913276	A	C	646	321	325	0.503	BRCA2	ENSG00000139618	NON_SYNONYMOUS_CODING	ENST00000380152.3:c.4784A>C	ENSP00000369497.3:p.Gln1595Pro	11/27	-	-	-	
S37	SNV	chr13	32913276	A	C	646	321	325	0.503	BRCA2	ENSG00000139618	NON_SYNONYMOUS_CODING	ENST00000544455.1:c.4784A>C	ENSP00000439902.1:p.Gln1595Pro	11/28	YES	-	-	
S59	INDEL	chr13	32903619	A	-TAC	533	331	202	0.379	BRCA2	ENSG00000139618	CODON_CHANGE_PLUS_CODON_DELETION	ENST00000380152.3:c.673delA	ENSP00000369497.3:p.Thr225LeufsTer5	8/27	-	-	-	
S59	INDEL	chr13	32903619	A	-TAC	533	331	202	0.379	BRCA2	ENSG00000139618	CODON_CHANGE_PLUS_CODON_DELETION	ENST00000544455.1:c.673delA	ENSP00000439902.1:p.Thr225LeufsTer5	8/28	YES	-	-	
S59	INDEL	chr13	32903619	A	-TAC	533	331	202	0.379	BRCA2	ENSG00000139618	CODON_CHANGE_PLUS_CODON_DELETION	ENST00000530893.2:c.304delA	ENSP00000435699.2:p.Thr102LeufsTer5	8/10	-	-	-	
S61	SNV	chr13	32912787	G	A	526	263	263	0.5	BRCA2	ENSG00000139618	NON_SYNONYMOUS_CODING	ENST00000380152.3:c.4295G>A	ENSP00000369497.3:p.Ser1432Asn	11/27	-	-	-	
S61	SNV	chr13	32912787	G	A	526	263	263	0.5	BRCA2	ENSG00000139618	NON_SYNONYMOUS_CODING	ENST00000544455.1:c.4295G>A	ENSP00000439902.1:p.Ser1432Asn	11/28	YES	-	-	
12	SNV	chr9	136910527	C	T	457	234	223	0.488	BRD3	ENSG00000169925	NON_SYNONYMOUS_CODING	ENST00000357885.2:c.1103G>A	ENSP00000350557.2:p.Arg368Gln	7/10	-	-	-	
12	SNV	chr9	136910527	C	T	457	234	223	0.488	BRD3	ENSG00000169925	NON_SYNONYMOUS_CODING	ENST00000371834.2:c.1103G>A	ENSP00000360900.2:p.Arg368Gln	7/10	-	-	-	
12	SNV	chr9	136910527	C	T	457	234	223	0.488	BRD3	ENSG00000169925	NON_SYNONYMOUS_CODING	ENST00000303407.7:c.1103G>A	ENSP00000305918.6:p.Arg368Gln	7/12	YES	-	-	
59	SNV	chr17	59761264	G	A	41	20	21	0.512	BRIP1	ENSG00000136492	NON_SYNONYMOUS_CODING	ENST00000259008.2:c.3143C>T	ENSP00000259008.2:p.Pro1048Leu	20/20	YES	-	-	

S44	INDEL	chr17	59876564	C	-T	698	353	345	0.494	BRIP1	ENSG00000136492	FRAME_SHIFT	ENST00000577598.1:c.1237delG	ENSP00000464654.1:p.Val413PhefsTer10	8/18	-	-	-
S44	INDEL	chr17	59876564	C	-T	698	353	345	0.494	BRIP1	ENSG00000136492	FRAME_SHIFT	ENST00000259008.2:c.1237delG	ENSP00000259008.2:p.Val413PhefsTer10	9/20	YES	-	-
S40	SNV	chr15	55710731	G	A	417	203	214	0.513	C15orf65	ENSG00000261652	NON_SYNONYMOUS_CODING	ENST00000569691.1:c.329G>A	ENSP00000456337.1:p.Arg110Lys	2/2	YES	-	-
46	INDEL	chr20	31040147	C	+CTGGGAGTAGGAG GAGGCTGG	157	119	38	0.239	C20orf112	ENSG00000197183	CODON_INSERTION	-	-	7/8	YES	-	-
S06	SNV	chr20	31052855	G	A	449	223	226	0.503	C20orf112	ENSG00000197183	NON_SYNONYMOUS_CODING	ENST00000375677.1:c.241C>T	ENSP00000364829.1:p.Arg81Trp	3/3	-	-	rs527734660
S06	SNV	chr20	31052855	G	A	449	223	226	0.503	C20orf112	ENSG00000197183	NON_SYNONYMOUS_CODING	ENST00000326071.4:c.175C>T	ENSP00000317413.4:p.Arg59Trp	3/3	-	-	rs527734660
S42	SNV	chr20	31041557	G	T	383	206	177	0.462	C20orf112	ENSG00000197183	NON_SYNONYMOUS_CODING	ENST00000359676.5:c.395C>A	ENSP00000352704.5:p.Pro132His	4/8	YES	0.0004	rs563159052
S62	SNV	chr20	31052855	G	A	675	330	345	0.51	C20orf112	ENSG00000197183	NON_SYNONYMOUS_CODING	ENST00000375677.1:c.241C>T	ENSP00000364829.1:p.Arg81Trp	3/3	-	-	rs527734660
S62	SNV	chr20	31052855	G	A	675	330	345	0.51	C20orf112	ENSG00000197183	NON_SYNONYMOUS_CODING	ENST00000326071.4:c.175C>T	ENSP00000317413.4:p.Arg59Trp	3/3	-	-	rs527734660
S47	SNV	chr2	24261989	C	T	660	329	331	0.501	C2orf44	ENSG00000163026	NON_SYNONYMOUS_CODING	ENST00000406895.3:c.376G>A	ENSP00000385816.3:p.Ala126Thr	2/3	-	-	-
S47	SNV	chr2	24261989	C	T	660	329	331	0.501	C2orf44	ENSG00000163026	NON_SYNONYMOUS_CODING	ENST00000295148.4:c.376G>A	ENSP00000295148.4:p.Ala126Thr	2/4	YES	-	-
S47	SNV	chr2	24261989	C	T	660	329	331	0.501	C2orf44	ENSG00000163026	NON_SYNONYMOUS_CODING	ENST00000443232.1:c.376G>A	ENSP00000413426.1:p.Ala126Thr	2/2	-	-	-
35	SNV	chr3	10123017	T	C	485	259	226	0.466	C3orf24	ENSG00000163705	UTR_3_PRIME	ENST00000524279.1:c.*242A>G	-	3/3	-	-	-
S03	SNV	chr1	7723761	C	T	489	247	242	0.495	CAMTA1	ENSG00000171735	NON_SYNONYMOUS_CODING	ENST00000303635.7:c.1154C>T	ENSP00000306522.6:p.Ala385Val	9/23	YES	-	COSM43046
S03	SNV	chr1	7723761	C	T	489	247	242	0.495	CAMTA1	ENSG00000171735	NON_SYNONYMOUS_CODING	ENST00000439411.2:c.1154C>T	ENSP00000402561.2:p.Ala385Val	9/22	-	-	COSM43046
35	SNV	chr17	76993226	T	A	512	275	237	0.463	CANT1	ENSG00000171302	NON_SYNONYMOUS_CODING	ENST00000302345.2:c.479A>T	ENSP00000307674.2:p.Glu160Val	2/4	YES	-	-
35	SNV	chr17	76993226	T	A	512	275	237	0.463	CANT1	ENSG00000171302	NON_SYNONYMOUS_CODING	ENST00000392446.5:c.479A>T	ENSP00000376241.4:p.Glu160Val	3/5	-	-	-
S05	SNV	chr7	2998203	G	A	170	89	81	0.476	CARD11	ENSG00000198286	START_GAINED	ENST00000356408.3:c.-63C>T	-	3/4	-	0.0002	rs573345021
S05	SNV	chr7	2998203	G	A	170	89	81	0.476	CARD11	ENSG00000198286	START_GAINED	ENST00000396946.4:c.-63C>T	-	2/25	YES	0.0002	rs573345021
S30	SNV	chr7	2998203	G	A	383	214	169	0.441	CARD11	ENSG00000198286	START_GAINED	ENST00000396946.4:c.-63C>T	-	2/25	YES	0.0002	rs573345021
S30	SNV	chr7	2998203	G	A	383	214	169	0.441	CARD11	ENSG00000198286	START_GAINED	ENST00000356408.3:c.-63C>T	-	3/4	-	0.0002	rs573345021
S18	SNV	chr11	3060480	G	C	292	135	157	0.538	CARS	ENSG00000110619	NON_SYNONYMOUS_CODING	ENST00000380525.4:c.595C>G	ENSP00000369897.4:p.Arg199Gly	6/23	YES	0.0002	rs555551728
S18	SNV	chr11	3060480	G	C	292	135	157	0.538	CARS	ENSG00000110619	NON_SYNONYMOUS_CODING	ENST00000278224.9:c.346C>G	ENSP00000278224.9:p.Arg116Gly	5/22	-	0.0002	rs555551728
S18	SNV	chr11	3060480	G	C	292	135	157	0.538	CARS	ENSG00000110619	NON_SYNONYMOUS_CODING	ENST00000401769.3:c.385C>G	ENSP00000384069.3:p.Arg129Gly	6/23	-	0.0002	rs555551728
S18	SNV	chr11	3060480	G	C	292	135	157	0.538	CARS	ENSG00000110619	NON_SYNONYMOUS_CODING	ENST00000397111.5:c.346C>G	ENSP00000380300.5:p.Arg116Gly	5/22	-	0.0002	rs555551728
S18	SNV	chr11	3060480	G	C	292	135	157	0.538	CARS	ENSG00000110619	NON_SYNONYMOUS_CODING	ENST00000397114.3:c.316C>G	ENSP00000380303.3:p.Arg106Gly	6/23	-	0.0002	rs555551728
S22	SNV	chr16	67070577	G	T	700	261	439	0.627	CBFB	ENSG00000067955	NON_SYNONYMOUS_CODING	ENST00000290858.6:c.201G>T	ENSP00000290858.6:p.Gln67His	3/6	-	-	-
S22	SNV	chr16	67070577	G	T	700	261	439	0.627	CBFB	ENSG00000067955	NON_SYNONYMOUS_CODING	ENST00000412916.2:c.201G>T	ENSP00000415151.2:p.Gln67His	3/6	YES	-	-
S51	SNV	chr16	67100648	C	T	591	307	284	0.479	CBFB	ENSG00000067955	NON_SYNONYMOUS_CODING	ENST00000564034.1:c.229C>T	ENSP00000456637.1:p.Leu77Phe	3/3	-	-	-
S51	SNV	chr16	67100648	C	T	591	307	284	0.479	CBFB	ENSG00000067955	NON_SYNONYMOUS_CODING	ENST00000561924.2:c.46C>T	ENSP00000462273.1:p.Leu16Phe	4/6	-	-	-
S51	SNV	chr16	67100648	C	T	591	307	284	0.479	CBFB	ENSG00000067955	NON_SYNONYMOUS_CODING	ENST00000290858.6:c.346C>T	ENSP00000290858.6:p.Leu116Phe	4/6	-	-	-
S51	SNV	chr16	67100648	C	T	591	307	284	0.479	CBFB	ENSG00000067955	NON_SYNONYMOUS_CODING	ENST00000412916.2:c.346C>T	ENSP00000415151.2:p.Leu116Phe	4/6	YES	-	-
S51	SNV	chr16	67100648	C	T	591	307	284	0.479	CBFB	ENSG00000067955	NON_SYNONYMOUS_CODING	ENST00000563939.2:c.46C>T	ENSP00000462148.1:p.Leu16Phe	3/4	-	-	-
21	SNV	chr11	119148891	T	C	161	43	118	0.733	CBL	ENSG00000110395	NON_SYNONYMOUS_CODING	ENST00000264033.4:c.1111T>C	ENSP00000264033.3:p.Tyr371His	8/16	YES	-	rs267606706,COSM34053,COSM34052,CM105459,CM099544
31	SNV	chr11	119148931	G	A	217	146	71	0.327	CBL	ENSG00000110395	NON_SYNONYMOUS_CODING	ENST00000264033.4:c.1151G>A	ENSP00000264033.3:p.Cys384Tyr	8/16	YES	-	COSM34066
43	INDEL	chr11	119148880	A	-ATATGAATT	191	146	45	0.234	CBL	ENSG00000110395	CODON_CHANGE_PLUS_CODON_DELETION	ENST00000264033.4:c.1101delA	ENSP00000264033.3:p.Gln367HisfsTer25	8/16	YES	-	COSM3444158,COSM1351837
58	SNV	chr11	119149260	T	A	249	65	184	0.739	CBL	ENSG00000110395	NON_SYNONYMOUS_CODING	ENST00000264033.4:c.1268T>A	ENSP00000264033.3:p.Ile423Asn	9/16	YES	-	-
S62	SNV	chr11	119149401	T	C	602	318	284	0.472	CBL	ENSG00000110395	NON_SYNONYMOUS_CODING	ENST00000264033.4:c.1121T>C	ENSP00000264033.3:p.Met374Thr	8/16	YES	-	-
S62	INDEL	chr10	61666069	C	-CCGCCGCCA	211	154	57	0.27	CCDC6	ENSG00000108091	CODON_DELETION	ENST00000263102.6:c.116delG	ENSP00000263102.6:p.Gly39GlufsTer35	1/9	YES	-	-
33	SNV	chr14	20781696	T	C	373	216	157	0.421	CONB1IP1	ENSG00000100814	NON_SYNONYMOUS_CODING	ENST00000437553.2:c.562A>G	ENSP00000409896.2:p.Ile188Val	6/7	-	-	-
33	SNV	chr14	20781696	T	C	373	216	157	0.421	CONB1IP1	ENSG00000100814	NON_SYNONYMOUS_CODING	ENST00000398160.2:c.562A>G	ENSP00000381226.2:p.Ile188Val	6/7	-	-	-
33	SNV	chr14	20781696	T	C	373	216	157	0.421	CONB1IP1	ENSG00000100814	NON_SYNONYMOUS_CODING	ENST00000398163.2:c.562A>G	ENSP00000381229.2:p.Ile188Val	6/7	-	-	-
33	SNV	chr14	20781696	T	C	373	216	157	0.421	CONB1IP1	ENSG00000100814	NON_SYNONYMOUS_CODING	ENST00000398169.3:c.562A>G	ENSP00000381235.3:p.Ile188Val	6/7	YES	-	-
33	SNV	chr14	20781696	T	C	373	216	157	0.421	CONB1IP1	ENSG00000100814	NON_SYNONYMOUS_CODING	ENST00000358932.4:c.562A>G	ENSP00000351810.4:p.Ile188Val	6/7	-	-	-
33	SNV	chr14	20781696	T	C	373	216	157	0.421	CONB1IP1	ENSG00000100814	NON_SYNONYMOUS_CODING	ENST00000553689.4:c.562A>G	ENSP00000337396.4:p.Ile188Val	5/6	YES	-	-
11	SNV	chr6	41908288	G	C	88	50	38	0.432	CCND3	ENSG00000112576	UTR_5_PRIME	ENST00000511942.1:c.-10C>G	-	2/5	-	-	-
11	SNV	chr6	41908288	G	C	88	50	38	0.432	CCND3	ENSG00000112576	NON_SYNONYMOUS_CODING	ENST00000514588.1:c.84C>G	ENSP00000420991.1:p.Phe28Leu	3/3	-	-	-
11	SNV	chr6	41908288	G	C	88	50	38	0.432	CCND3	ENSG00000112576	UTR_5_PRIME	ENST00000502771.1:c.-10C>G	-	3/3	-	-	-
11	SNV	chr6	41908288	G	C	88	50	38	0.432	CCND3	ENSG00000112576	UTR_5_PRIME	ENST00000510503.1:c.-10C>G	-	2/4	-	-	-
11	SNV	chr6	41908288	G	C	88	50	38	0.432	CCND3	ENSG00000112576	UTR_5_PRIME	ENST00000372988.4:c.-10C>G	-	2/5	-	-	-
11	SNV	chr6	41908288	G	C	88	50	38	0.432	CCND3	ENSG00000112576	UTR_5_PRIME	ENST00000505064.1:c.-10C>G	-	1/3	-	-	-
11	SNV	chr6	41908288	G	C	88	50	38	0.432	CCND3	ENSG00000112576	NON_SYNONYMOUS_CODING	ENST00000372987.4:c.84C>G	ENSP00000362078.4:p.Phe28Leu	2/5	-	-	-
11	SNV	chr6	41908288	G	C	88	50	38	0.432	CCND3	ENSG00000112576	NON_SYNONYMOUS_CODING	ENST00000372991.4:c.234C>G	ENSP00000362082.4:p.Phe78Leu	2/5	YES	-	-
S36	INDEL	chr19	30308100	C	-AAA	894	488	406	0.454	CCNE1	ENSG00000105173	CODON_CHANGE_PLUS_CODON_DELETION	ENST00000444983.2:c.193delA	ENSP00000410179.2:p.Glu66LysfsTer28	3/10	-	-	-
S36	INDEL	chr19	30308100	C	-AAA	894	488	406	0.454	CCNE1	ENSG00000105173	CODON_CHANGE_PLUS_CODON_DELETION	ENST00000357943.5:c.238delA	ENSP00000350625.5:p.Glu81LysfsTer28	5/11	-	-	-
S36	INDEL	chr19	30308100	C	-AAA	894	488	406	0.454	CCNE1	ENSG00000105173	CODON_CHANGE_PLUS_CODON_DELETION	ENST00000575243.1:c.229delA	ENSP00000459024.1:p.Glu78LysfsTer28	5/6	-	-	-
S36	INDEL	chr19	30308100	C	-AAA	894	488	406	0.454	CCNE1	ENSG00000105173	CODON_CHANGE_PLUS_CODON_DELETION	ENST00000262643.3:c.238delA	ENSP00000262643.3:p.Glu81LysfsTer28	5/12	YES	-	-
S18	INDEL	chr22	37964375	G	CTGCAAACCCCCCA GCCACTA	127	88	39	0.307	CDC42EP1	ENSG00000128283	CODON_DELETION	ENST00000249014.4:c.725delC	ENSP00000249014.4:p.Ala242ValfsTer49	3/3	YES	-	-
33	SNV	chr1	193205402	G	A	311	170	141	0.453	CDC73	ENSG00000134371	NON_SYNONYMOUS_CODING	ENST00000367435.3:c.1333G>A	ENSP00000356405.3:p.Val445Ile	15/17	YES	-	rs368442389,COSM901401
35	SNV	chr16	64981819	C	T	304	230	74	0.243	CDH11	ENSG00000140937	UTR_3_PRIME	ENST00000394156.3:c.*175G>A	-	14/14	-	-	COSM1200408
35	SNV	chr16	64981819	C	T	304	230	74	0.243	CDH11	ENSG00000140937	NON_SYNONYMOUS_CODING	ENST00000268603.4:c.2078G>A	ENSP00000268603.4:p.Arg693His	13/13	YES	-	COSM1200408
1	INDEL	chr13	28542772	G	-TGCGGGTGGTGA	37	25	12	0.324	GDX2	ENSG00000165556	CODON_DELETION	ENST00000381020.7:c.373delC	ENSP00000370408.6:p.His125ThrfsTer57	1/3	YES	-	-
S18	INDEL	chr19	33792661	C	+A	32	17	15	0.469	CEBPA	ENSG00000245848	FRAME_SHIFT	-	-	1/1	YES	-	-
S26	INDEL	chr19	33792731	G	+GCGGGT	103	76	27	0.262	CEBPA	ENSG00000245848	CODON_CHANGE_PLUS_CODON_INSERTION	-	-	1/1	YES	-	-
S32	SNV	chr19	33792447	T	C	796	400	396	0.498	CEBPA	ENSG00000245848	NON_SYNONYMOUS_CODING	ENST00000498907.2:c.874A>G	ENSP00000427514.1:p.Asn292Asp	1/1	YES	-	-
S38	INDEL	chr19	33793053	T	+A	274	215	59	0.215	CEBPA	ENSG00000245848	FRAME_SHIFT	-	-	1/1	YES	-	-
S61	INDEL	chr19	33792512	C	+CG	700	391	309	0.441	CEBPA	ENSG00000245848	FRAME_SHIFT	-	-	1/1	YES	-	-
48	SNV	chr1	22307647	G	A	223	122	101	0.453	CELA3B	ENSG00000219073	NON_SYNONYMOUS_CODING	ENST00000400277.2:c.53G>A	ENSP00000383135.2:p.Arg18His	1/5	-	-	-
48	SNV	chr1	22307647	G	A	223	122	101	0.453	CELA3B	ENSG00000219073	NON_SYNONYMOUS_CODING	ENST00000337107.6:c.344G>A	ENSP00000338369.6:p.Arg115His	4/8	YES	-	-
48	SNV	chr1	22307647	G	A	223	122	101	0.453	CELA3B	ENSG00000219073	NON_SYNONYMOUS_CODING	ENST00000374666.1:c.392G>A	ENSP00000363798.1:p.Arg131His	4/5	-	-	-
S29	SNV	chr1	22213034	G	C	165	80	85	0.515	CELA3B	ENSG00000219073	NON_SYNONYMOUS_CODING	ENST00000337107.6:c.653G>C	ENSP00000338369.6:p.Gly218Ala	7/8	YES	-	-
S29	SNV	chr1	22213034	G	C	165	80	85	0.515	CELA3B	ENSG00000219073	NON_SYNONYMOUS_CODING	ENST00000400277.2:c.362G>C	ENSP00000383135.2:p.Gly121Ala	4/5	-	-	-
32	SNV	chr22	29121087	A	G	131	76	55	0.42	CHEK2	ENSG00000183765	NON_SYNONYMOUS_CODING	ENST00000402731.1:c.470T>C	ENSP00000384835.1:p.Ile157Thr	3/13	-	0.0010	rs17879961,COSM3693990,CM993368
32	SNV	chr22	29121087	A	G	131	76	55	0.42	CHEK2	ENSG00000183765	NON_SYNONYMOUS_CODING	ENST00000439200.1:c.563T>C	ENSP00000408065.1:p.Ile188Thr	5/7	-	0.0010	rs17879961,COSM3693990,CM993368
32	SNV	chr22	29121087	A	G	131	76	55	0.42	CHEK2	ENSG00000183765	START_GAINED	ENST00000544772.1:c.-308T>C	-	4/16	-	0.0010	rs17879961,COSM3693990,CM993368
32	SNV	chr22	29121087	A	G	131	76	55	0.42	CHEK2	ENSG00000183765	NON_SYNONYMOUS_CODING	ENST00000348295.3:c.470T>C	ENSP00000329012.5:p.Ile157Thr	4/14	-	0.0010	rs17879961,COSM3693990,CM993368
32	SNV	chr22	29121087	A	G	131	76	55	0.42	CHEK2	ENSG00000183765	NON_SYNONYMOUS_CODING	ENST00000328354.6:c.470T>C	ENSP00000329178.6:p.Ile157Thr	4/15	-	0.0010	rs17879961,COSM3693990,CM993368
32	SNV	chr22	29121087	A	G	131	76	55	0.42	CHEK2	ENSG00000183765	NON_SYNONYMOUS_CODING	ENST00000382566.1:c.470T>C	ENSP00000372023.2:p.Ile157Thr	4/14	-	0.0010	rs17879961,COSM3693990,CM993368
32	SNV	chr22	29121087	A	G	131	76	55	0.42	CHEK2	ENSG00000183765	NON_SYNONYMOUS_CODING	ENST00000382580.2:c.599T>C	ENSP00000372023.2:p.Ile200Thr	5/16	YES	0.0010	rs17879961,COSM3693990,CM993368
32	SNV	chr22	29121087	A	G	131	76	55	0.42	CHEK2	ENSG00000183765	NON_SYNONYMOUS_CODING	ENST00000398017.2:c.500T>C	ENSP00000381099.2:p.Ile167Thr	6/6	-	0.0010	rs17879961,COSM3693990,CM993368
32	SNV	chr22	29121087	A	G	131	76	55	0.42	CHEK2	ENSG00000183765	NON_SYNONYMOUS_CODING	ENST00000405598.1:c.470T>C	ENSP00000386087.1:p.Ile157Thr	5/16	-	0.0010	rs17879961,COSM3693990,CM993368
32	SNV	chr22	29121087	A	G	131	76	55	0.42	CHEK2	ENSG00000183765	UTR_5_PRIME	ENST00000425190.2:c.-194T>C	-	3/9	-	0.0010	rs17879961,COSM3693990,CM993368
32	SNV	chr22	29121087	A	G	131	76	55	0.42	CHEK2	ENSG00000183765	NON_SYNONYMOUS_CODING	ENST00000404276.1:c.470T>C	ENSP00000385747.1:p.Ile157Thr	3/14	-	0.0010	rs17879961,COSM3693990,CM993368
35	SNV	chr22	29121087	A	G	361	196	165	0.457	CHEK2	ENSG00000183765	UTR_5_PRIME	ENST00000425190.2:c.-194T>C	-	3/9	-	0.0010	rs17879961,COSM3693990,CM993368
35	SNV	chr22	29121087	A	G	361	196	165	0.457	CHEK2	ENSG00000183765	NON_SYNONYMOUS_CODING	ENST00000439200.1:c.563T>C	ENSP00000408065.1:p.Ile188Thr	5/7	-	0.0010	rs17879961,COSM3693990,CM993368
35	SNV	chr22	29121087	A	G	361	196	165	0.457	CHEK2	ENSG00000183765	NON_SYNONYMOUS_CODING	ENST00000328354.6:c.470T>C	ENSP00000329178.6:p.Ile157Thr	4/15	-	0.0010	rs17879961,COSM3693990,CM993368
35	SNV	chr22	29121087	A	G	361	196	165	0.457	CHEK2	ENSG00000183765	NON_SYNONYMOUS_CODING	ENST00000382580.2:c.599T>C	ENSP00000372023.2:p.Ile200Thr	5/16	YES	0.0010	rs17879961,COSM3693990,CM993368
35	SNV	chr22	29121087	A	G	361	196	165	0.457	CHEK2	ENSG00000183765	NON_SYNONYMOUS_CODING	ENST00000402731.1:c.470T>C	ENSP00000384835.1:p.Ile157Thr	3/13	-	0.0010	rs17879961,COSM3693990,CM993368
35	SNV	chr22	29121087	A	G	361	196	165	0.457	CHEK2	ENSG00000183765	NON_SYNONYMOUS_CODING	ENST00000404276.1:c.470T>C	ENSP00000385747.1:p.Ile157Thr	3/14	-	0.0010	rs17879961,COSM3693990,CM993368
35	SNV	chr22	29121087	A	G	361	196	165	0.457	CHEK2	ENSG00000183765	START_GAINED	ENST00000544772.1:c.-308T>C	-	4/16	-	0.0010	rs17879961,COSM3693990,CM993368
35	SNV	chr22	29121087	A	G	361	196	165	0.457	CHEK2	ENSG00000183765	NON_SYNONYMOUS_CODING	ENST00000348295.3:c.470T>C	ENSP00000329012.5:p.Ile157Thr	4/14	-	0.0010	rs17879961,COSM3693990,CM993368
35	SNV	chr22	29121087	A	G	361	196	165	0.457	CHEK2	ENSG00000183765	NON_SYNONYMOUS_CODING	ENST00000382566.1:c.470T>C	ENSP00000372023.2:p.Ile157Thr	4/14	-	0.0010	rs17879961,COSM3693990,CM993368
35	SNV	chr22	29121087	A	G	361	196	165	0.457	CHEK2	ENSG00000183765	NON_SYNONYMOUS_CODING	ENST00000398017.2:c.500T>C	ENSP00000381099.2:p.Ile167Thr	6/6	-	0.0010	rs17879961,COSM3693990,CM993368
35	SNV	chr22	29121087	A	G	361	196	165	0.457	CHEK2	ENSG00000183765	NON_SYNONYMOUS_CODING	ENST00000405598.1:c.470T>C	ENSP00000386087.1:p.Ile157Thr	5/16	-	0.0010	rs17879961,COSM3693990,CM993368
38	INDEL	chr22	29091856	A	-G	473	241	232	0.49	CHEK2	ENSG00000183765	FRAME_SHIFT	ENST00000403642.1:c.828delT	ENSP00000384919.1:p.Asp277IlefsTer14	8/12	-	0.0010	rs555607708,KinMutBase_CHEK2_DNA:g.49841delC,CD993415
38	INDEL	chr22	29091856	A	-G	473	241	232	0.49	CHEK2	ENSG00000183765	FRAME_SHIFT	ENST00000382580.2:c.1230delT	ENSP00000372023.2:p.Asp411IlefsTer14	12/16	YES	0.0010	rs555607708,KinMutBase_CHEK2_DNA:g.49841delC,CD993415

38	INDEL	chr22	29091856	A	-G	473	241	232	0.49	CHEK2	ENSG00000183765	FRAME_SHIFT	ENST00000544772.1:c.438delT	ENSP00000442458.1:p.Asp147IlefsTer14	12/16	-	0.0010	rs555607708,KinMutBase_CHEK2_DNA:g.49841delC,CD993415
38	INDEL	chr22	29091856	A	-G	473	241	232	0.49	CHEK2	ENSG00000183765	FRAME_SHIFT	ENST00000402731.1:c.1014delT	ENSP00000384835.1:p.Asp339IlefsTer14	9/13	-	0.0010	rs555607708,KinMutBase_CHEK2_DNA:g.49841delC,CD993415
38	INDEL	chr22	29091856	A	-G	473	241	232	0.49	CHEK2	ENSG00000183765	UTR_3_PRIME	ENST00000382566.1:c.*169delT		10/14	-	0.0010	rs555607708,KinMutBase_CHEK2_DNA:g.49841delC,CD993415
38	INDEL	chr22	29091856	A	-G	473	241	232	0.49	CHEK2	ENSG00000183765	FRAME_SHIFT	ENST00000382578.1:c.828delT	ENSP00000372021.1:p.Asp277IlefsTer14	9/13	-	0.0010	rs555607708,KinMutBase_CHEK2_DNA:g.49841delC,CD993415
38	INDEL	chr22	29091856	A	-G	473	241	232	0.49	CHEK2	ENSG00000183765	FRAME_SHIFT	ENST00000405598.1:c.1101delT	ENSP00000386087.1:p.Asp368IlefsTer14	12/16	-	0.0010	rs555607708,KinMutBase_CHEK2_DNA:g.49841delC,CD993415
38	INDEL	chr22	29091856	A	-G	473	241	232	0.49	CHEK2	ENSG00000183765	FRAME_SHIFT	ENST00000348295.3:c.1014delT	ENSP00000329012.5:p.Asp339IlefsTer14	10/14	-	0.0010	rs555607708,KinMutBase_CHEK2_DNA:g.49841delC,CD993415
38	INDEL	chr22	29091856	A	-G	473	241	232	0.49	CHEK2	ENSG00000183765	FRAME_SHIFT	ENST00000328354.6:c.1101delT	ENSP00000329178.6:p.Asp368IlefsTer14	11/15	-	0.0010	rs555607708,KinMutBase_CHEK2_DNA:g.49841delC,CD993415
38	INDEL	chr22	29091856	A	-G	473	241	232	0.49	CHEK2	ENSG00000183765	FRAME_SHIFT	ENST00000404276.1:c.1101delT	ENSP00000385747.1:p.Asp368IlefsTer14	10/14	-	0.0010	rs555607708,KinMutBase_CHEK2_DNA:g.49841delC,CD993415
43	SNV	chr22	29121087	A	G	331	172	159	0.48	CHEK2	ENSG00000183765	NON_SYNONYMOUS_CODING	ENST00000398017.2:c.500T>C	ENSP00000381099.2:p.Ile167Thr	6/6	-	0.0010	rs17879961,COSM3693990,CM993368
43	SNV	chr22	29121087	A	G	331	172	159	0.48	CHEK2	ENSG00000183765	NON_SYNONYMOUS_CODING	ENST00000328354.6:c.470T>C	ENSP00000329178.6:p.Ile157Thr	4/15	-	0.0010	rs17879961,COSM3693990,CM993368
43	SNV	chr22	29121087	A	G	331	172	159	0.48	CHEK2	ENSG00000183765	NON_SYNONYMOUS_CODING	ENST00000402731.1:c.470T>C	ENSP00000384835.1:p.Ile157Thr	3/13	-	0.0010	rs17879961,COSM3693990,CM993368
43	SNV	chr22	29121087	A	G	331	172	159	0.48	CHEK2	ENSG00000183765	NON_SYNONYMOUS_CODING	ENST00000382566.1:c.470T>C	ENSP00000372007.1:p.Ile157Thr	4/14	-	0.0010	rs17879961,COSM3693990,CM993368
43	SNV	chr22	29121087	A	G	331	172	159	0.48	CHEK2	ENSG00000183765	NON_SYNONYMOUS_CODING	ENST00000404276.1:c.470T>C	ENSP00000385747.1:p.Ile157Thr	3/14	-	0.0010	rs17879961,COSM3693990,CM993368
43	SNV	chr22	29121087	A	G	331	172	159	0.48	CHEK2	ENSG00000183765	NON_SYNONYMOUS_CODING	ENST00000439200.1:c.563T>C	ENSP00000408065.1:p.Ile188Thr	5/7	-	0.0010	rs17879961,COSM3693990,CM993368
43	SNV	chr22	29121087	A	G	331	172	159	0.48	CHEK2	ENSG00000183765	UTR_5_PRIME	ENST00000425190.2:c.-194T>C	-	3/9	-	0.0010	rs17879961,COSM3693990,CM993368
43	SNV	chr22	29121087	A	G	331	172	159	0.48	CHEK2	ENSG00000183765	NON_SYNONYMOUS_CODING	ENST00000382580.2:c.599T>C	ENSP00000372023.2:p.Ile200Thr	5/16	YES	0.0010	rs17879961,COSM3693990,CM993368
43	SNV	chr22	29121087	A	G	331	172	159	0.48	CHEK2	ENSG00000183765	NON_SYNONYMOUS_CODING	ENST00000405598.1:c.470T>C	ENSP00000386087.1:p.Ile157Thr	5/16	-	0.0010	rs17879961,COSM3693990,CM993368
43	SNV	chr22	29121087	A	G	331	172	159	0.48	CHEK2	ENSG00000183765	START_GAINED	ENST00000544772.1:c.-308T>C	-	4/16	-	0.0010	rs17879961,COSM3693990,CM993368
43	SNV	chr22	29121087	A	G	331	172	159	0.48	CHEK2	ENSG00000183765	NON_SYNONYMOUS_CODING	ENST00000348295.3:c.470T>C	ENSP00000329012.5:p.Ile157Thr	4/14	-	0.0010	rs17879961,COSM3693990,CM993368
S02	SNV	chr22	29121087	A	G	439	216	223	0.508	CHEK2	ENSG00000183765	NON_SYNONYMOUS_CODING	ENST00000382580.2:c.599T>C	ENSP00000372023.2:p.Ile200Thr	5/16	YES	0.0010	rs17879961,COSM3693990,CM993368
S02	SNV	chr22	29121087	A	G	439	216	223	0.508	CHEK2	ENSG00000183765	START_GAINED	ENST00000544772.1:c.-308T>C	-	4/16	-	0.0010	rs17879961,COSM3693990,CM993368
S02	SNV	chr22	29121087	A	G	439	216	223	0.508	CHEK2	ENSG00000183765	NON_SYNONYMOUS_CODING	ENST00000398017.2:c.500T>C	ENSP00000381099.2:p.Ile167Thr	6/6	-	0.0010	rs17879961,COSM3693990,CM993368
S02	SNV	chr22	29121087	A	G	439	216	223	0.508	CHEK2	ENSG00000183765	NON_SYNONYMOUS_CODING	ENST00000439200.1:c.563T>C	ENSP00000408065.1:p.Ile188Thr	5/7	-	0.0010	rs17879961,COSM3693990,CM993368
S02	SNV	chr22	29121087	A	G	439	216	223	0.508	CHEK2	ENSG00000183765	START_GAINED	ENST00000425190.2:c.-194T>C	-	3/9	-	0.0010	rs17879961,COSM3693990,CM993368
S02	SNV	chr22	29121087	A	G	439	216	223	0.508	CHEK2	ENSG00000183765	NON_SYNONYMOUS_CODING	ENST00000328354.6:c.470T>C	ENSP00000329178.6:p.Ile157Thr	4/15	-	0.0010	rs17879961,COSM3693990,CM993368
S02	SNV	chr22	29121087	A	G	439	216	223	0.508	CHEK2	ENSG00000183765	NON_SYNONYMOUS_CODING	ENST00000382566.1:c.470T>C	ENSP00000372007.1:p.Ile157Thr	4/14	-	0.0010	rs17879961,COSM3693990,CM993368
S02	SNV	chr22	29121087	A	G	439	216	223	0.508	CHEK2	ENSG00000183765	NON_SYNONYMOUS_CODING	ENST00000348295.3:c.470T>C	ENSP00000329012.5:p.Ile157Thr	4/14	-	0.0010	rs17879961,COSM3693990,CM993368
S02	SNV	chr22	29121087	A	G	439	216	223	0.508	CHEK2	ENSG00000183765	NON_SYNONYMOUS_CODING	ENST00000405598.1:c.470T>C	ENSP00000386087.1:p.Ile157Thr	5/16	-	0.0010	rs17879961,COSM3693990,CM993368
S02	SNV	chr22	29121087	A	G	439	216	223	0.508	CHEK2	ENSG00000183765	NON_SYNONYMOUS_CODING	ENST00000402731.1:c.470T>C	ENSP00000384835.1:p.Ile157Thr	3/13	-	0.0010	rs17879961,COSM3693990,CM993368
S02	SNV	chr22	29121087	A	G	439	216	223	0.508	CHEK2	ENSG00000183765	NON_SYNONYMOUS_CODING	ENST00000404276.1:c.470T>C	ENSP00000385747.1:p.Ile157Thr	3/14	-	0.0010	rs17879961,COSM3693990,CM993368
S59	SNV	chr22	29121087	A	G	684	357	327	0.478	CHEK2	ENSG00000183765	NON_SYNONYMOUS_CODING	ENST00000398017.2:c.500T>C	ENSP00000381099.2:p.Ile167Thr	6/6	-	0.0010	rs17879961,COSM3693990,CM993368
S59	SNV	chr22	29121087	A	G	684	357	327	0.478	CHEK2	ENSG00000183765	NON_SYNONYMOUS_CODING	ENST00000328354.6:c.470T>C	ENSP00000329178.6:p.Ile157Thr	4/15	-	0.0010	rs17879961,COSM3693990,CM993368
S59	SNV	chr22	29121087	A	G	684	357	327	0.478	CHEK2	ENSG00000183765	NON_SYNONYMOUS_CODING	ENST00000382580.2:c.599T>C	ENSP00000372023.2:p.Ile200Thr	5/16	YES	0.0010	rs17879961,COSM3693990,CM993368
S59	SNV	chr22	29121087	A	G	684	357	327	0.478	CHEK2	ENSG00000183765	START_GAINED	ENST00000425190.2:c.-194T>C	-	3/9	-	0.0010	rs17879961,COSM3693990,CM993368
S59	SNV	chr22	29121087	A	G	684	357	327	0.478	CHEK2	ENSG00000183765	NON_SYNONYMOUS_CODING	ENST00000402731.1:c.470T>C	ENSP00000384835.1:p.Ile157Thr	3/13	-	0.0010	rs17879961,COSM3693990,CM993368
S59	SNV	chr22	29121087	A	G	684	357	327	0.478	CHEK2	ENSG00000183765	NON_SYNONYMOUS_CODING	ENST00000382566.1:c.470T>C	ENSP00000372007.1:p.Ile157Thr	4/14	-	0.0010	rs17879961,COSM3693990,CM993368
S59	SNV	chr22	29121087	A	G	684	357	327	0.478	CHEK2	ENSG00000183765	NON_SYNONYMOUS_CODING	ENST00000348295.3:c.470T>C	ENSP00000329012.5:p.Ile157Thr	4/14	-	0.0010	rs17879961,COSM3693990,CM993368
S59	SNV	chr22	29121087	A	G	684	357	327	0.478	CHEK2	ENSG00000183765	NON_SYNONYMOUS_CODING	ENST00000404276.1:c.470T>C	ENSP00000385747.1:p.Ile157Thr	3/14	-	0.0010	rs17879961,COSM3693990,CM993368
S59	SNV	chr22	29121087	A	G	684	357	327	0.478	CHEK2	ENSG00000183765	START_GAINED	ENST00000544772.1:c.-308T>C	-	4/16	-	0.0010	rs17879961,COSM3693990,CM993368
S59	SNV	chr22	29121087	A	G	684	357	327	0.478	CHEK2	ENSG00000183765	NON_SYNONYMOUS_CODING	ENST00000405598.1:c.470T>C	ENSP00000386087.1:p.Ile157Thr	5/16	-	0.0010	rs17879961,COSM3693990,CM993368
S59	SNV	chr22	29121087	A	G	684	357	327	0.478	CHEK2	ENSG00000183765	NON_SYNONYMOUS_CODING	ENST00000439200.1:c.563T>C	ENSP00000408065.1:p.Ile188Thr	5/7	-	0.0010	rs17879961,COSM3693990,CM993368
26	SNV	chr2	175711883	A	C	121	70	51	0.422	CHN1	ENSG00000128656	UTR_5_PRIME	ENST00000444573.1:c.-24T>G	-	1/4	-	-	-
26	SNV	chr2	175711883	A	C	121	70	51	0.422	CHN1	ENSG00000128656	UTR_5_PRIME	ENST00000295497.7:c.-24T>G	-	1/7	-	-	-
24	SNV	chr19	42797928	C	T	57	31	26	0.456	CIC	ENSG00000079432	NON_SYNONYMOUS_CODING	ENST00000160740.3:c.3974C>T	ENSP00000160740.3:p.Thr1325Ile	16/20	-	-	-
S15	SNV	chr19	42799134	C	A	133	82	51	0.384	CIC	ENSG00000079432	NON_SYNONYMOUS_CODING	ENST00000575354.2:c.4618C>A	ENSP00000458663.2:p.Pro1540Thr	20/20	YES	-	-
S15	SNV	chr19	42799134	C	A	133	82	51	0.384	CIC	ENSG00000079432	NON_SYNONYMOUS_CODING	ENST00000576505.2:c.752C>A	ENSP00000459011.2:p.Pro252Thr	5/5	-	-	-
S15	SNV	chr19	42799134	C	A	133	82	51	0.384	CIC	ENSG00000079432	NON_SYNONYMOUS_CODING	ENST00000572681.2:c.7336C>A	ENSP00000459719.1:p.Pro2446Thr	21/21	-	-	-
S15	SNV	chr19	42799134	C	A	133	82	51	0.384	CIC	ENSG00000079432	NON_SYNONYMOUS_CODING	ENST00000160740.3:c.4612C>A	ENSP00000160740.3:p.Pro1538Thr	20/20	-	-	-
S15	SNV	chr19	42799134	C	A	133	82	51	0.384	CIC	ENSG00000079432	NON_SYNONYMOUS_CODING	ENST00000573349.1:c.799C>A	ENSP00000458543.1:p.Pro267Thr	5/5	-	-	-
S35	SNV	chr19	42776512	A	T	276	143	133	0.482	CIC	ENSG00000079432	NON_SYNONYMOUS_CODING	ENST00000572681.2:c.577A>T	ENSP00000459719.1:p.Thr193Ser	2/21	-	-	rs547565552
S42	SNV	chr19	42795740	C	G	334	158	176	0.527	CIC	ENSG00000079432	NON_SYNONYMOUS_CODING	ENST00000160740.3:c.2729C>G	ENSP00000160740.3:p.Pro910Arg	11/20	-	-	-
S42	SNV	chr19	42795740	C	G	334	158	176	0.527	CIC	ENSG00000079432	NON_SYNONYMOUS_CODING	ENST00000575354.2:c.2729C>G	ENSP00000458663.2:p.Pro910Arg	11/20	YES	-	-
S42	SNV	chr19	42795740	C	G	334	158	176	0.527	CIC	ENSG00000079432	NON_SYNONYMOUS_CODING	ENST00000572681.2:c.5456C>G	ENSP00000459719.1:p.Pro1819Arg	12/21	-	-	-
S48	SNV	chr19	42777790	G	A	933	334	599	0.642	CIC	ENSG00000079432	NON_SYNONYMOUS_CODING	ENST00000572681.2:c.1855G>A	ENSP00000459719.1:p.Val619Ile	2/21	-	-	-
S54	SNV	chr19	42776512	A	T	496	263	233	0.47	CIC	ENSG00000079432	NON_SYNONYMOUS_CODING	ENST00000572681.2:c.577A>T	ENSP00000459719.1:p.Thr193Ser	2/21	-	-	rs547565552
S30	SNV	chr16	11001448	G	A	401	214	187	0.466	CIITA	ENSG00000179583	NON_SYNONYMOUS_CODING	ENST00000324288.8:c.2099G>A	ENSP00000316328.8:p.Arg700Gln	11/20	YES	-	-
S56	SNV	chr16	11016073	G	A	712	419	293	0.412	CIITA	ENSG00000179583	NON_SYNONYMOUS_CODING	ENST00000381835.5:c.1447G>A	ENSP00000371257.5:p.Val483Met	15/18	-	-	-
S56	SNV	chr16	11016073	G	A	712	419	293	0.412	CIITA	ENSG00000179583	NON_SYNONYMOUS_CODING	ENST00000324288.8:c.3199G>A	ENSP00000316328.8:p.Val1067Met	17/20	YES	-	-
S19	SNV	chr22	19263296	A	G	601	275	326	0.542	CLTCL1	ENSG00000070371	NON_SYNONYMOUS_CODING	ENST00000353891.5:c.100T>C	ENSP00000439662.1:p.Ser34Pro	2/32	-	-	-
S19	SNV	chr22	19263296	A	G	601	275	326	0.542	CLTCL1	ENSG00000070371	NON_SYNONYMOUS_CODING	ENST00000263200.10:c.100T>C	ENSP00000445677.1:p.Ser34Pro	2/33	YES	-	-
S19	SNV	chr22	19263296	A	G	601	275	326	0.542	CLTCL1	ENSG00000070371	NON_SYNONYMOUS_CODING	ENST00000449918.1:c.100T>C	ENSP00000443264.1:p.Ser34Pro	2/4	-	-	-
S19	SNV	chr22	19263296	A	G	601	275	326	0.542	CLTCL1	ENSG00000070371	NON_SYNONYMOUS_CODING	ENST00000427926.1:c.100T>C	ENSP00000441158.1:p.Ser34Pro	2/34	-	-	-
S61	SNV	chr22	19220991	T	A	710	343	367	0.516	CLTCL1	ENSG00000070371	NON_SYNONYMOUS_CODING	ENST00000427926.1:c.1322A>T	ENSP00000441158.1:p.Gln441Leu	8/34	-	-	-
S61	SNV	chr22	19220991	T	A	710	343	367	0.516	CLTCL1	ENSG00000070371	NON_SYNONYMOUS_CODING	ENST00000353891.5:c.1322A>T	ENSP00000439662.1:p.Gln441Leu	8/32	-	-	-
S61	SNV	chr22	19220991	T	A	710	343	367	0.516	CLTCL1	ENSG00000070371	NON_SYNONYMOUS_CODING	ENST00000263200.10:c.1322A>T	ENSP00000445677.1:p.Gln441Leu	8/33	YES	-	-
63	SNV	chr17	48278940	C	T	73	39	34	0.466	COL1A1	ENSG00000108821	UTR_5_PRIME	ENST00000225964.5:c.-66G>A	-	1/51	YES	0.0002	rs565562045
63	SNV	chr17	48270375	G	C	232	122	110	0.474	COL1A1	ENSG00000108821	NON_SYNONYMOUS_CODING	ENST00000225964.5:c.1801C>G	ENSP00000225964.5:p.Pro601Ala	26/51	YES	-	-
25	SNV	chr2	208440093	T	G	347	201	146	0.421	CREB1	ENSG00000118260	UTR_3_PRIME	ENST00000539789.1:c.*188T>G	-	7/9	-	-	-
55	SNV	chr7	137686502	G	C	75	40	35	0.467	CREB3L2	ENSG00000182158	UTR_5_PRIME	ENST00000330387.6:c.-51C>G	-	1/12	YES	-	-
55	SNV	chr7	137686502	G	C	75	40	35	0.467	CREB3L2	ENSG00000182158	UTR_5_PRIME	ENST00000452463.1:c.-51C>G	-	1/4	-	-	-
55	SNV	chr7	137686502	G	C	75	40	35	0.467	CREB3L2	ENSG00000182158	UTR_5_PRIME	ENST00000456390.1:c.-51C>G	-	1/10	-	-	-
6	SNV	chr16	3820668	G	A	362	190	172	0.475	CREBBP	ENSG00000005339	NON_SYNONYMOUS_CODING	ENST00000382070.3:c.2669C>T	ENSP00000371502.3:p.Pro890Leu	13/30	-	-	-
6	SNV	chr16	3820668	G	A	362	190	172	0.475	CREBBP	ENSG00000005339	NON_SYNONYMOUS_CODING	ENST00000262367.5:c.2783C>T	ENSP00000262367.5:p.Pro928Leu	14/31	YES	-	-
S46	SNV	chr16	3828111	G	A	358	267	91	0.254	CREBBP	ENSG00000005339	NON_SYNONYMOUS_CODING	ENST00000572134.1:c.327C>T	ENSP00000458254.1:p.Arg110Cys	3/5	-	-	COSM175791
S46	SNV	chr16	3828111	G	A	358	267	91	0.254	CREBBP	ENSG00000005339	NON_SYNONYMOUS_CODING	ENST00000262367.5:c.2014C>T	ENSP00000262367.5:p.Arg672Cys	10/31	YES	-	COSM175791
S46	SNV	chr16	3828111	G	A	358	267	91	0.254	CREBBP	ENSG00000005339	NON_SYNONYMOUS_CODING	ENST00000571826.1:c.634C>T	ENSP00000459490.1:p.Arg212Cys	1/4	-	-	COSM175791
S46	SNV	chr16	3828111	G	A	358	267	91	0.254	CREBBP	ENSG00000005339	NON_SYNONYMOUS_CODING	ENST00000382070.3:c.1900C>T	ENSP00000371502.3:p.Arg634Cys	9/30	-	-	COSM175791
46	SNV	chr19	18885731	G	A	425	220	205	0.482	CRTC1	ENSG00000105662	NON_SYNONYMOUS_CODING	ENST00000338797.6:c.1495G>A	ENSP00000345001.5:p.Val499Met	13/15	YES	-	-
46	SNV	chr19	18885731	G	A	425	220	205	0.482	CRTC1	ENSG00000105662	NON_SYNONYMOUS_CODING	ENST00000321949.8:c.1447G>A	ENSP00000323332.7:p.Val483Met	12/14	-	-	-
S14	SNV	chr19	18885735	T	C	166	106	60	0.361	CRTC1	ENSG00000105662	NON_SYNONYMOUS_CODING	ENST00000321949.8:c.1451T>C	ENSP00000323332.7:p.Phe484Ser	12/14	-	-	-
S14	SNV	chr19	18885735	T	C	166	106	60	0.361	CRTC1	ENSG00000105662	NON_SYNONYMOUS_CODING	ENST00000594658.1:c.1328T>C	ENSP00000468893.1:p.Phe443Ser	12/14	-	-	-
S14	SNV	chr19	18885735	T	C	166	106	60	0.361	CRTC1	ENSG00000105662	NON_SYNONYMOUS_CODING	ENST00000338797.6:c.1499T>C	ENSP00000345001.5:p.Phe500Ser	13/15	YES	-	-
S26	SNV	chr15	91172643	C	A	297	125	172	0.579	CRTC3	ENSG00000140577	NON_SYNONYMOUS_CODING	ENST00000268184.6:c.1145C>A	ENSP00000268184.6:p.Pro382Gln	11/15	YES	-	-
S26	SNV	chr15	91172643	C	A	297	125	172	0.579	CRTC3	ENSG00000140577	NON_SYNONYMOUS_CODING	ENST00000420329.2:c.1145C>A	ENSP00000416573.2:p.Pro382Gln	11/15	-	-	-
26	SNV	chr16	50813611	T	G	331	179	152	0.459	CYLD	ENSG00000083799	NON_SYNONYMOUS_CODING	ENST00000398568.2:c.1165T>G	ENSP00000381574.2:p.Ser389Ala	8/18	-	-	-
26	SNV	chr16	50813611	T	G	331	179	152	0.459	CYLD	ENSG00000083799	NON_SYNONYMOUS_CODING	ENST00000311559.9:c.1174T>G	ENSP00000308928.9:p.Ser392Ala	10/20	-	-	-
26	SNV	chr16	50813611	T	G	331	179	152	0.459	CYLD	ENSG00000083799	NON_SYNONYMOUS_CODING	ENST00000427738.3:c.1174T>G	ENSP00000392025.3:p.Ser392Ala	8/18	YES	-	-
26	SNV	chr16	50813611	T	G	331	179	152	0.459	CYLD	ENSG00000083799	NON_SYNONYMOUS_CODING	ENST00000540145.1:c.1174T>G	ENSP00000445447.1:p.Ser392Ala	9/19	-	-	-
S59	SNV	chr16	50783697	G	A	514	257	257	0.5	CYLD	ENSG00000083799	NON_SYNONYMOUS_CODING	ENST00000566679.2:c.88G>A	ENSP00000456912.2:p.Val30Ile	3/5	-	-	-
S59	SNV	chr16	50783697	G	A	514	257	257	0.5	CYLD	ENSG00000083799	NON_SYNONYMOUS_CODING	ENST00000569418.1:c.88G>A	ENSP00000457576.1:p.Val30Ile	3/18	-	-	-
S59	SNV	chr16	50783697	G	A	514	257	257	0.5	CYLD	ENSG00000083799	NON_SYNONYMOUS_CODING	ENST00000540145.1:c.88G>A	ENSP00000445447.1:p.Val30Ile	4/19	-	-	-
S59	SNV	chr16	50783697	G	A	514	257	257	0.5	CYLD	ENSG00000083799	NON_SYNONYMOUS_CODING	ENST00000568704.2:c.88G>A	ENSP00000456488.2:p.Val30Ile	1/14	-	-	-
S59	SNV	chr16	50783697	G	A	514	257	257	0.5	CYLD	ENSG00000083799	NON_SYNONYMOUS_CODING	ENST00000563426.1:c.88G>A	ENSP00000454515.1:p.Val30Ile	2/17	-	-	-
S59	SNV	chr16	50783697	G	A	514	257	257	0.5	CYLD	ENSG00000083799	NON_SYNONYMOUS_CODING	ENST00000427738.3:c.88G>A	ENSP00000392025.3:p.Val30Ile	2/18	YES	-	-
S59	SNV	chr16	50783697	G	A	514	257	257	0.5	CYLD	ENSG00000083799	NON_SYNONYMOUS_CODING	ENST00000398568.2:c.88G>A	ENSP00000381574.2:p.Val30Ile	3/18	-	-	-
S59	SNV	chr16	50783697	G	A	514	257	257	0.5	CYLD	ENSG00000083799	NON_SYNONYMOUS_CODING	ENST00000569681.1:c.88G>A	ENSP00000457108.1:p.Val30Ile	3/3	-	-	-
S59	SNV	chr16	50783697	G	A	514	257	257	0.5	CYLD	ENSG00000083799	NON_SYNONYMOUS_CODING	ENST00000564634.1:c.88G>A	ENSP00000455535.1:p.Val30Ile	4/4	-	-	-
S59	SNV	chr16	50783697	G	A	514	257	257	0.5	CYLD	ENSG00000083799	NON_SYNONYMOUS_CODING	ENST00000311559.9:c.88G>A	ENSP00000308928.9:p.Val30Ile	4/20	-	-	-
S59	SNV	chr16	50783697	G	A	514	257	257	0.5	CYLD	ENSG00000083799	NON_SYNONYMOUS_CODING	ENST00000566206.1:c.88G>A	ENSP00000462134.1:p.Val30Ile	3/18	-	-	-

62	SNV	chr9	124535601	C	G	50	28	22	0.44	DAB2IP	ENSG00000136848	NON_SYNONYMOUS_CODING	ENST00000373782.3:c.2521C>G	ENSP00000362887.3:p.Arg841Gly	10/15	-	-	-
62	SNV	chr9	124535601	C	G	50	28	22	0.44	DAB2IP	ENSG00000136848	NON_SYNONYMOUS_CODING	ENST00000259371.2:c.2710C>G	ENSP00000259371.2:p.Arg904Gly	12/17	YES	-	-
62	SNV	chr9	124535601	C	G	50	28	22	0.44	DAB2IP	ENSG00000136848	NON_SYNONYMOUS_CODING	ENST00000408936.3:c.2794C>G	ENSP00000386183.3:p.Arg932Gly	12/16	-	-	-
62	SNV	chr9	124535601	C	G	50	28	22	0.44	DAB2IP	ENSG00000136848	NON_SYNONYMOUS_CODING	ENST00000309989.1:c.2422C>G	ENSP00000310827.1:p.Arg808Gly	10/14	-	-	-
S03	SNV	chr9	124414086	C	T	48	25	23	0.479	DAB2IP	ENSG00000136848	NON_SYNONYMOUS_CODING	ENST00000408936.3:c.32C>T	ENSP00000386183.3:p.Thr11Ile	1/16	-	0.0036	rs576970717
S21	SNV	chr13	72049882	G	A	476	233	243	0.51	DACH1	ENSG00000165659	NON_SYNONYMOUS_CODING	ENST00000313174.7:c.1532C>T	ENSP00000318506.7:p.Thr511Met	6/8	-	-	-
S21	SNV	chr13	72049882	G	A	476	233	243	0.51	DACH1	ENSG00000165659	NON_SYNONYMOUS_CODING	ENST00000359684.2:c.2132C>T	ENSP00000352712.2:p.Thr711Met	10/12	-	-	-
S21	SNV	chr13	72049882	G	A	476	233	243	0.51	DACH1	ENSG00000165659	NON_SYNONYMOUS_CODING	ENST00000305425.4:c.1976C>T	ENSP00000304994.4:p.Thr659Met	9/11	YES	-	-
S21	SNV	chr13	72049882	G	A	476	233	243	0.51	DACH1	ENSG00000165659	NON_SYNONYMOUS_CODING	ENST00000354591.4:c.1370C>T	ENSP00000346604.4:p.Thr457Met	5/7	-	-	-
S64	SNV	chr13	72063217	T	C	843	454	389	0.461	DACH1	ENSG00000165659	NON_SYNONYMOUS_CODING	ENST00000359684.2:c.1796A>G	ENSP00000352712.2:p.His599Arg	8/12	-	-	-
S64	SNV	chr13	72063217	T	C	843	454	389	0.461	DACH1	ENSG00000165659	NON_SYNONYMOUS_CODING	ENST00000354591.4:c.1034A>G	ENSP00000346604.4:p.His345Arg	3/7	-	-	-
S64	SNV	chr13	72063217	T	C	843	454	389	0.461	DACH1	ENSG00000165659	NON_SYNONYMOUS_CODING	ENST00000305425.4:c.1640A>G	ENSP00000304994.4:p.His547Arg	7/11	YES	-	-
S64	SNV	chr13	72063217	T	C	843	454	389	0.461	DACH1	ENSG00000165659	NON_SYNONYMOUS_CODING	ENST00000313174.7:c.1196A>G	ENSP00000318506.7:p.His399Arg	4/8	-	-	-
S43	SNV	chr11	126213205	G	A	667	239	428	0.642	DCPS	ENSG00000110063	NON_SYNONYMOUS_CODING	ENST00000263579.4:c.640G>A	ENSP00000263579.4:p.Asp214Asn	5/6	YES	-	-
55	SNV	chr17	62496694	G	T	321	177	144	0.449	DDX5	ENSG00000108654	NON_SYNONYMOUS_CODING	ENST00000450599.2:c.1177C>A	ENSP00000403085.2:p.Leu393Ile	11/12	-	-	-
55	SNV	chr17	62496694	G	T	321	177	144	0.449	DDX5	ENSG00000108654	NON_SYNONYMOUS_CODING	ENST00000225792.5:c.1414C>A	ENSP00000225792.5:p.Leu472Ile	12/13	YES	-	-
S44	SNV	chr17	62496288	C	A	735	388	347	0.472	DDX5	ENSG00000108654	NON_SYNONYMOUS_CODING	ENST00000225792.5:c.1598G>T	ENSP00000225792.5:p.Gly533Val	13/13	YES	-	COSM74350
S44	SNV	chr17	62496288	C	A	735	388	347	0.472	DDX5	ENSG00000108654	NON_SYNONYMOUS_CODING	ENST00000450599.2:c.1361G>T	ENSP00000403085.2:p.Gly454Val	12/12	-	-	COSM74350
S44	SNV	chr17	62496288	C	A	735	388	347	0.472	DDX5	ENSG00000108654	NON_SYNONYMOUS_CODING	ENST00000578804.1:c.1598G>T	ENSP00000462885.1:p.Gly533Val	13/13	-	-	COSM74350
S54	SNV	chr17	62501974	C	A	589	291	298	0.506	DDX5	ENSG00000108654	START_GAINED	ENST00000578804.1:c.-67G>T	-	1/13	-	0.0002	rs566114785
S46	SNV	chr6	18255982	G	T	437	218	219	0.501	DEK	ENSG00000124795	NON_SYNONYMOUS_CODING	ENST00000244776.7:c.451C>A	ENSP00000244776.7:p.His151Asn	5/10	-	-	-
S46	SNV	chr6	18255982	G	T	437	218	219	0.501	DEK	ENSG00000124795	NON_SYNONYMOUS_CODING	ENST00000397239.3:c.553C>A	ENSP00000380414.3:p.His185Asn	6/11	YES	-	-
S46	SNV	chr6	18255982	G	T	437	218	219	0.501	DEK	ENSG00000124795	NON_SYNONYMOUS_CODING	ENST00000503715.1:c.352C>A	ENSP00000425399.1:p.His118Asn	5/5	-	-	-
S54	INDEL	chr19	10940881	G	+C	362	252	110	0.304	DNM2	ENSG00000079805	FRAME_SHIFT	-	-	20/21	-	-	COSM1390225,COSM1390224
S54	INDEL	chr19	10940881	G	+C	362	252	110	0.304	DNM2	ENSG00000079805	FRAME_SHIFT	-	-	20/21	YES	-	COSM1390225,COSM1390224
S54	INDEL	chr19	10940881	G	+C	362	252	110	0.304	DNM2	ENSG00000079805	FRAME_SHIFT	-	-	2/2	-	-	COSM1390225,COSM1390224
S54	INDEL	chr19	10940881	G	+C	362	252	110	0.304	DNM2	ENSG00000079805	FRAME_SHIFT	-	-	20/21	-	-	COSM1390225,COSM1390224
S54	INDEL	chr19	10940881	G	+C	362	252	110	0.304	DNM2	ENSG00000079805	FRAME_SHIFT	-	-	19/20	-	-	COSM1390225,COSM1390224
S54	INDEL	chr19	10940881	G	+C	362	252	110	0.304	DNM2	ENSG00000079805	FRAME_SHIFT	-	-	20/21	-	-	COSM1390225,COSM1390224
S54	INDEL	chr19	10940881	G	+C	362	252	110	0.304	DNM2	ENSG00000079805	FRAME_SHIFT	-	-	19/20	-	-	COSM1390225,COSM1390224
S26	SNV	chr19	10248653	A	G	109	46	63	0.578	DNMT1	ENSG00000130816	NON_SYNONYMOUS_CODING	ENST00000540357.1:c.4100T>C	ENSP00000440457.1:p.Val1367Ala	35/40	-	-	-
S26	SNV	chr19	10248653	A	G	109	46	63	0.578	DNMT1	ENSG00000130816	NON_SYNONYMOUS_CODING	ENST00000340748.4:c.4100T>C	ENSP00000345739.3:p.Val1367Ala	35/40	-	-	-
S26	SNV	chr19	10248653	A	G	109	46	63	0.578	DNMT1	ENSG00000130816	NON_SYNONYMOUS_CODING	ENST00000359526.4:c.4148T>C	ENSP00000352516.3:p.Val1383Ala	36/41	YES	-	-
11	SNV	chr2	25457242	C	T	188	114	74	0.394	DNMT3A	ENSG00000119772	NON_SYNONYMOUS_CODING	ENST00000402667.1:c.1976G>A	ENSP00000384237.1:p.Arg659His	18/18	-	-	rs147001633,COSM99740,COSM52944,COSM1583129,COSM3356083,COSM442676
11	SNV	chr2	25457242	C	T	188	114	74	0.394	DNMT3A	ENSG00000119772	NON_SYNONYMOUS_CODING	ENST00000321117.5:c.2645G>A	ENSP00000324375.5:p.Arg882His	23/23	-	-	rs147001633,COSM99740,COSM52944,COSM1583129,COSM3356083,COSM442676
11	SNV	chr2	25457242	C	T	188	114	74	0.394	DNMT3A	ENSG00000119772	NON_SYNONYMOUS_CODING	ENST00000380746.4:c.2078G>A	ENSP00000370122.4:p.Arg693His	19/19	-	-	rs147001633,COSM99740,COSM52944,COSM1583129,COSM3356083,COSM442676
11	SNV	chr2	25457242	C	T	188	114	74	0.394	DNMT3A	ENSG00000119772	NON_SYNONYMOUS_CODING	ENST00000264709.3:c.2645G>A	ENSP00000264709.3:p.Arg882His	23/23	YES	-	rs147001633,COSM99740,COSM52944,COSM1583129,COSM3356083,COSM442676
20	SNV	chr2	25457243	G	A	329	177	152	0.462	DNMT3A	ENSG00000119772	NON_SYNONYMOUS_CODING	ENST00000380746.4:c.2077C>T	ENSP00000370122.4:p.Arg693Cys	19/19	-	-	rs377577594,COSM1583136,COSM87001,COSM53042,COSM1166704
20	SNV	chr2	25457243	G	A	329	177	152	0.462	DNMT3A	ENSG00000119772	NON_SYNONYMOUS_CODING	ENST00000264709.3:c.2644C>T	ENSP00000264709.3:p.Arg882Cys	23/23	YES	-	rs377577594,COSM1583136,COSM87001,COSM53042,COSM1166704
20	SNV	chr2	25457243	G	A	329	177	152	0.462	DNMT3A	ENSG00000119772	NON_SYNONYMOUS_CODING	ENST00000402667.1:c.1975C>T	ENSP00000384237.1:p.Arg659Cys	18/18	-	-	rs377577594,COSM1583136,COSM87001,COSM53042,COSM1166704
20	SNV	chr2	25457243	G	A	329	177	152	0.462	DNMT3A	ENSG00000119772	NON_SYNONYMOUS_CODING	ENST00000321117.5:c.2644C>T	ENSP00000324375.5:p.Arg882Cys	23/23	-	-	rs377577594,COSM1583136,COSM87001,COSM53042,COSM1166704
21	INDEL	chr2	25463235	C	-AGA	240	54	186	0.775	DNMT3A	ENSG00000119772	CODON_CHANGE_PLUS_CODON_DELETION	ENST00000380746.4:c.1692delG	ENSP00000370122.4:p.Trp564CysfsTer26	15/19	-	-	COSM133134
21	INDEL	chr2	25463235	C	-AGA	240	54	186	0.775	DNMT3A	ENSG00000119772	CODON_CHANGE_PLUS_CODON_DELETION	ENST00000264709.3:c.2259delG	ENSP00000264709.3:p.Trp753CysfsTer26	19/23	YES	-	COSM133134
21	INDEL	chr2	25463235	C	-AGA	240	54	186	0.775	DNMT3A	ENSG00000119772	CODON_CHANGE_PLUS_CODON_DELETION	ENST00000402667.1:c.1590delG	ENSP00000384237.1:p.Trp530CysfsTer26	14/18	-	-	COSM133134
21	INDEL	chr2	25463235	C	-AGA	240	54	186	0.775	DNMT3A	ENSG00000119772	CODON_CHANGE_PLUS_CODON_DELETION	ENST00000321117.5:c.2259delG	ENSP00000324375.5:p.Trp753CysfsTer26	19/23	-	-	COSM133134
23	SNV	chr2	25457249	T	C	250	30	220	0.88	DNMT3A	ENSG00000119772	NON_SYNONYMOUS_CODING	ENST00000402667.1:c.1969A>G	ENSP00000384237.1:p.Met657Val	18/18	-	-	COSM120499
23	SNV	chr2	25457249	T	C	250	30	220	0.88	DNMT3A	ENSG00000119772	NON_SYNONYMOUS_CODING	ENST00000380746.4:c.2071A>G	ENSP00000370122.4:p.Met691Val	19/19	-	-	COSM120499
23	SNV	chr2	25457249	T	C	250	30	220	0.88	DNMT3A	ENSG00000119772	NON_SYNONYMOUS_CODING	ENST00000321117.5:c.2638A>G	ENSP00000324375.5:p.Met880Val	23/23	-	-	COSM120499
23	SNV	chr2	25457249	T	C	250	30	220	0.88	DNMT3A	ENSG00000119772	NON_SYNONYMOUS_CODING	ENST00000264709.3:c.2638A>G	ENSP00000264709.3:p.Met880Val	23/23	YES	-	COSM120499
24	SNV	chr2	25463248	G	A	215	123	92	0.428	DNMT3A	ENSG00000119772	NON_SYNONYMOUS_CODING	ENST00000380746.4:c.1678C>T	ENSP00000370122.4:p.Arg560Cys	15/19	-	-	COSM219133,COSM3580252
24	SNV	chr2	25463248	G	A	215	123	92	0.428	DNMT3A	ENSG00000119772	NON_SYNONYMOUS_CODING	ENST00000402667.1:c.1576C>T	ENSP00000384237.1:p.Arg526Cys	14/18	-	-	COSM219133,COSM3580252
24	SNV	chr2	25463248	G	A	215	123	92	0.428	DNMT3A	ENSG00000119772	NON_SYNONYMOUS_CODING	ENST00000321117.5:c.2245C>T	ENSP00000324375.5:p.Arg749Cys	19/23	-	-	COSM219133,COSM3580252
24	SNV	chr2	25463248	G	A	215	123	92	0.428	DNMT3A	ENSG00000119772	NON_SYNONYMOUS_CODING	ENST00000264709.3:c.2245C>T	ENSP00000264709.3:p.Arg749Cys	19/23	YES	-	COSM219133,COSM3580252
31	SNV	chr2	25463228	A	C	197	19	178	0.904	DNMT3A	ENSG00000119772	NON_SYNONYMOUS_CODING	ENST00000380746.4:c.1698T>G	ENSP00000370122.4:p.Phe566Leu	15/19	-	-	-
31	SNV	chr2	25463228	A	C	197	19	178	0.904	DNMT3A	ENSG00000119772	NON_SYNONYMOUS_CODING	ENST00000321117.5:c.2265T>G	ENSP00000324375.5:p.Phe755Leu	19/23	-	-	-
31	SNV	chr2	25463228	A	C	197	19	178	0.904	DNMT3A	ENSG00000119772	NON_SYNONYMOUS_CODING	ENST00000402667.1:c.1596T>G	ENSP00000384237.1:p.Phe532Leu	14/18	-	-	-
31	SNV	chr2	25463228	A	C	197	19	178	0.904	DNMT3A	ENSG00000119772	NON_SYNONYMOUS_CODING	ENST00000264709.3:c.2265T>G	ENSP00000264709.3:p.Phe755Leu	19/23	YES	-	-
40	SNV	chr2	25457243	G	A	147	87	60	0.408	DNMT3A	ENSG00000119772	NON_SYNONYMOUS_CODING	ENST00000402667.1:c.1975C>T	ENSP00000384237.1:p.Arg659Cys	18/18	-	-	rs377577594,COSM1583136,COSM87001,COSM53042,COSM1166704
40	SNV	chr2	25457243	G	A	147	87	60	0.408	DNMT3A	ENSG00000119772	NON_SYNONYMOUS_CODING	ENST00000321117.5:c.2644C>T	ENSP00000324375.5:p.Arg882Cys	23/23	-	-	rs377577594,COSM1583136,COSM87001,COSM53042,COSM1166704
40	SNV	chr2	25457243	G	A	147	87	60	0.408	DNMT3A	ENSG00000119772	NON_SYNONYMOUS_CODING	ENST00000264709.3:c.2644C>T	ENSP00000264709.3:p.Arg882Cys	23/23	YES	-	rs377577594,COSM1583136,COSM87001,COSM53042,COSM1166704
40	SNV	chr2	25457243	G	A	147	87	60	0.408	DNMT3A	ENSG00000119772	NON_SYNONYMOUS_CODING	ENST00000380746.4:c.2077C>T	ENSP00000370122.4:p.Arg693Cys	19/19	-	-	rs377577594,COSM1583136,COSM87001,COSM53042,COSM1166704
42	INDEL	chr2	25457181	G	-A	308	190	118	0.383	DNMT3A	ENSG00000119772	FRAME_SHIFT	ENST00000402667.1:c.2037delC	ENSP00000384237.1:p.Phe679LeufsTer4	18/18	-	-	rs587777510
42	INDEL	chr2	25457181	G	-A	308	190	118	0.383	DNMT3A	ENSG00000119772	FRAME_SHIFT	ENST00000264709.3:c.2706delC	ENSP00000264709.3:p.Phe902LeufsTer4	23/23	YES	-	rs587777510
42	INDEL	chr2	25457181	G	-A	308	190	118	0.383	DNMT3A	ENSG00000119772	FRAME_SHIFT	ENST00000321117.5:c.2706delC	ENSP00000324375.5:p.Phe902LeufsTer4	23/23	-	-	rs587777510
42	INDEL	chr2	25457181	G	-A	308	190	118	0.383	DNMT3A	ENSG00000119772	FRAME_SHIFT	ENST00000380746.4:c.2139delC	ENSP00000370122.4:p.Phe713LeufsTer4	19/19	-	-	rs587777510
42	SNV	chr2	25457183	A	G	309	190	119	0.385	DNMT3A	ENSG00000119772	NON_SYNONYMOUS_CODING	ENST00000321117.5:c.2704T>C	ENSP00000324375.5:p.Phe902Leu	23/23	-	-	-
42	SNV	chr2	25457183	A	G	309	190	119	0.385	DNMT3A	ENSG00000119772	NON_SYNONYMOUS_CODING	ENST00000380746.4:c.2137T>C	ENSP00000370122.4:p.Phe713Leu	19/19	-	-	-
42	SNV	chr2	25457183	A	G	309	190	119	0.385	DNMT3A	ENSG00000119772	NON_SYNONYMOUS_CODING	ENST00000264709.3:c.2704T>C	ENSP00000264709.3:p.Phe902Leu	23/23	YES	-	-
42	SNV	chr2	25457183	A	G	309	190	119	0.385	DNMT3A	ENSG00000119772	NON_SYNONYMOUS_CODING	ENST00000402667.1:c.2035T>C	ENSP00000384237.1:p.Phe679Leu	18/18	-	-	-
44	SNV	chr2	25462040	G	C	400	251	149	0.372	DNMT3A	ENSG00000119772	NON_SYNONYMOUS_CODING	ENST00000402667.1:c.1698C>G	ENSP00000384237.1:p.His566Gln	15/18	-	-	-
44	SNV	chr2	25462040	G	C	400	251	149	0.372	DNMT3A	ENSG00000119772	NON_SYNONYMOUS_CODING	ENST00000321117.5:c.2367C>G	ENSP00000324375.5:p.His789Gln	20/23	-	-	-
44	INDEL	chr2	25462028	G	+T	369	232	137	0.371	DNMT3A	ENSG00000119772	FRAME_SHIFT	-	-	20/23	YES	-	COSM231553
44	INDEL	chr2	25462028	G	+T	369	232	137	0.371	DNMT3A	ENSG00000119772	FRAME_SHIFT	-	-	16/19	-	-	COSM231553
44	INDEL	chr2	25462028	G	+T	369	232	137	0.371	DNMT3A	ENSG00000119772	FRAME_SHIFT	-	-	15/18	-	-	COSM231553
44	SNV	chr2	25462040	G	C	400	251	149	0.372	DNMT3A	ENSG00000119772	NON_SYNONYMOUS_CODING	ENST00000264709.3:c.2367C>G	ENSP00000264709.3:p.His789Gln	20/23	YES	-	-
44	SNV	chr2	25462040	G	C	400	251	149	0.372	DNMT3A	ENSG00000119772	NON_SYNONYMOUS_CODING	ENST00000380746.4:c.1800C>G	ENSP00000370122.4:p.His600Gln	16/19	-	-	-
44	INDEL	chr2	25462028	G	+T	369	232	137	0.371	DNMT3A	ENSG00000119772	FRAME_SHIFT	-	-	20/23	-	-	COSM231553
46	SNV	chr2	25457242	C	T	261	132	129	0.494	DNMT3A	ENSG00000119772	NON_SYNONYMOUS_CODING	ENST00000380746.4:c.2078G>A	ENSP00000370122.4:p.Arg693His	19/19	-	-	rs147001633,COSM99740,COSM52944,COSM1583129,COSM3356083,COSM442676
46	SNV	chr2	25457242	C	T	261	132	129	0.494	DNMT3A	ENSG00000119772	NON_SYNONYMOUS_CODING	ENST00000264709.3:c.2645G>A	ENSP00000264709.3:p.Arg882His	23/23	YES	-	rs147001633,COSM99740,COSM52944,COSM1583129,COSM3356083,COSM442676
46	SNV	chr2	25457242	C	T	261	132	129	0.494	DNMT3A	ENSG00000119772	NON_SYNONYMOUS_CODING	ENST00000402667.1:c.1976G>A	ENSP00000384237.1:p.Arg659His	18/18	-	-	rs147001633,COSM99740,COSM52944,COSM1583129,COSM3356083,COSM442676
46	SNV	chr2	25457242	C	T	261	132	129	0.494	DNMT3A	ENSG00000119772	NON_SYNONYMOUS_CODING	ENST00000321117.5:c.2645G>A	ENSP00000324375.5:p.Arg882His	23/23	-	-	rs147001633,COSM99740,COSM52944,COSM1583129,COSM3356083,COSM442676
58	INDEL	chr2	25469616	G	-AACAGCTTCCC	151	103	48	0.318	DNMT3A	ENSG00000119772	FRAME_SHIFT	ENST00000402667.1:c.485delC	ENSP00000384237.1:p.Pro162ArgfsTer22	5/18	-	-	-
58	INDEL	chr2	25469616	G	-AACAGCTTCCC	151	103	48	0.318	DNMT3A	ENSG00000119772	FRAME_SHIFT	ENST00000380746.4:c.587delC	ENSP00000370122.4:p.Pro196ArgfsTer22	6/19	-	-	-
58	SNV	chr2	25464537	C	G	256	139	117	0.457	DNMT3A	ENSG00000119772	NON_SYNONYMOUS_CODING	ENST00000321117.5:c.1976G>C	ENSP00000324375.5:p.Gly659Ala	17/23	-	-	COSM1019235,COSM1019236
58	INDEL	chr2	25469616	G	-AACAGCTTCCC	151	103	48	0.318	DNMT3A	ENSG00000119772	FRAME_SHIFT	ENST00000264709.3:c.1154delC	ENSP00000264709.3:p.Pro385ArgfsTer22	10/23	YES	-	-
58	SNV	chr2	25464537	C	G	256	139	117	0.457	DNMT3A	ENSG00000119772	NON_SYNONYMOUS_CODING	ENST00000264709.3:c.1976G>C	ENSP00000264709.3:p.Arg659Pro	17/23	YES	-	COSM1019235,COSM1019236
58	INDEL	chr2	25469616	G	-AACAGCTTCCC	151	103	48	0.318	DNMT3A	ENSG00000119772	FRAME_SHIFT	ENST00000321117.5:c.1154delC	ENSP00000324375.5:p.Pro385ArgfsTer22	10/23	-	-	-
58	SNV	chr2	25464537	C	G	256	139	117	0.457	DNMT3A	ENSG00000119772	NON_SYNONYMOUS_CODING	ENST00000402667.1:c.1307G>C	ENSP00000384237.1:p.Arg436Pro	12/18	-	-	COSM1019235,COSM1019236
58	SNV	chr2	25464537	C	G	256	139	117	0.457	DNMT3A	ENSG00000119772	NON_SYNONYMOUS_CODING	ENST00000380746.4:c.1409G>C	ENSP00000370122.4:p.Arg470Pro	13/19	-	-	COSM1019235,COSM1019236
59	INDEL	chr2	25467149	T	-GGCTGCCTG	169	84	85	0.503	DNMT3A	ENSG00000119772	CODON_CHANGE_PLUS_CODON_DELETION	ENST00000321117.5:c.1726delA	ENSP00000324375.5:p.Ile576LeufsTer75	15/23	-	-	-
59	INDEL	chr2	25467149	T	-GGCTGCCTG	169	84	85	0.503	DNMT3A	ENSG00000119772	CODON_CHANGE_PLUS_CODON_DELETION	ENST00000264709.3:c.1726delA	ENSP00000264709.3:p.Ile576LeufsTer75	15/23	YES	-	-
59	INDEL	chr2	25467149	T	-GGCTGCCTG	169	84	85	0.503	DNMT3A	ENSG00000119772	CODON_CHANGE_PLUS_CODON_DELETION	ENST00000380746.4:c.1159delA	ENSP00000370122.4:p.Ile387LeufsTer75	11/19	-	-	-
59	INDEL	chr2	25467149	T	-GGCTGCCTG	169	84	85	0.503	DNMT3A	ENSG00000119772	CODON_CHANGE_PLUS_CODON_DELETION	ENST00000402667.1:c.1057delA	ENSP00000384237.1:p.Ile353LeufsTer75	10/18	-	-	-
S02	SNV	chr2	25457243	G	A	325	181	144	0.443	DNMT3A	ENSG00000119772	NON_SYNONYMOUS_CODING	ENST00000264709.3:c.2644C>T	ENSP00000264709.3:p.Arg882Cys	23/23	YES	-	rs377577594,COSM1583136,COSM87001,COSM53042,COSM1166704
S02	SNV	chr2	25457243	G	A	325	181	144	0.443	DNMT3A	ENSG00000119772	NON_SYNONYMOUS_CODING	ENST00000321117.5:c.2644C>T	ENSP00000324375.5:p.Arg882Cys	23/23	-	-	rs377577594,COSM1583136,COSM87001,COSM53042,COSM1166704
S02	SNV	chr2	25457243	G	A	325	181	144	0.443	DNMT3A	ENSG00000119772	NON_SYNONYMOUS_CODING	ENST00000380746.4:c.2077C>T	ENSP00000370122.4:p.Arg693Cys	19/19	-	-	rs377577594,COSM1583136,COSM87001,COSM53042,COSM1166704
S02	SNV	chr2	25457243	G	A	325	181	144	0.443	DNMT3A	ENSG00000119772	NON_SYNONYMOUS_CODING	ENST00000402667.1:c.1975C>T	ENSP00000384237.1:p.Arg659Cys	18/18	-	-	rs377577594,COSM1583136,COSM87001,COSM53042,COSM1166704
S05	SNV	chr2	25467436	A	T	367	199	168	0.458	DNMT3A	ENSG00000119772	NON_SYNONYMOUS_CODING	ENST00000380746.4:c.1073T>A	ENSP00000370122.4:p.Leu358His	10/19	-	-	rs111848085,COSM231556,COSM1583072
S05	SNV	chr2	25467436	A	T	367	199	168	0.458	DNMT3A	ENSG00000119772	NON_SYNONYMOUS_CODING	ENST00000321117.5:c.1640T>A	ENSP00000324375.5:p.Leu547His	14/23	-	-	rs111848085,COSM231556,COSM1583072

S05	SNV	chr2	25467436	A	T	367	199	168	0.458	DNMT3A	ENSG00000119772	NON_SYNONYMOUS_CODING	ENST00000264709.3:c.1640T>A	ENSP00000264709.3:p.Leu547His	14/23	YES	-	rs111848085,COSM231556,COSM1583072
S05	SNV	chr2	25467436	A	T	367	199	168	0.458	DNMT3A	ENSG00000119772	NON_SYNONYMOUS_CODING	ENST00000402667.1:c.971T>A	ENSP00000384237.1:p.Leu324His	9/18	-	-	rs111848085,COSM231556,COSM1583072
S17	SNV	chr2	25457242	C	T	654	333	321	0.491	DNMT3A	ENSG00000119772	NON_SYNONYMOUS_CODING	ENST00000380746.4:c.2078G>A	ENSP00000370122.4:p.Arg693His	19/19	-	-	rs147001633,COSM9974O,COSM52944,COSM1583129,COSM3356083,COSM442676
S17	SNV	chr2	25457242	C	T	654	333	321	0.491	DNMT3A	ENSG00000119772	NON_SYNONYMOUS_CODING	ENST00000402667.1:c.1976G>A	ENSP00000384237.1:p.Arg659His	18/18	-	-	rs147001633,COSM9974O,COSM52944,COSM1583129,COSM3356083,COSM442676
S17	SNV	chr2	25457242	C	T	654	333	321	0.491	DNMT3A	ENSG00000119772	NON_SYNONYMOUS_CODING	ENST00000321117.5:c.2645G>A	ENSP00000324375.5:p.Arg882His	23/23	-	-	rs147001633,COSM9974O,COSM52944,COSM1583129,COSM3356083,COSM442676
S17	SNV	chr2	25457242	C	T	654	333	321	0.491	DNMT3A	ENSG00000119772	NON_SYNONYMOUS_CODING	ENST00000264709.3:c.2645G>A	ENSP00000264709.3:p.Arg882His	23/23	YES	-	rs147001633,COSM9974O,COSM52944,COSM1583129,COSM3356083,COSM442676
S19	SNV	chr2	25457242	C	T	491	271	220	0.448	DNMT3A	ENSG00000119772	NON_SYNONYMOUS_CODING	ENST00000380746.4:c.2078G>A	ENSP00000370122.4:p.Arg693His	19/19	-	-	rs147001633,COSM9974O,COSM52944,COSM1583129,COSM3356083,COSM442676
S19	SNV	chr2	25457242	C	T	491	271	220	0.448	DNMT3A	ENSG00000119772	NON_SYNONYMOUS_CODING	ENST00000402667.1:c.1976G>A	ENSP00000384237.1:p.Arg659His	18/18	-	-	rs147001633,COSM9974O,COSM52944,COSM1583129,COSM3356083,COSM442676
S19	SNV	chr2	25457242	C	T	491	271	220	0.448	DNMT3A	ENSG00000119772	NON_SYNONYMOUS_CODING	ENST00000264709.3:c.2645G>A	ENSP00000264709.3:p.Arg882His	23/23	YES	-	rs147001633,COSM9974O,COSM52944,COSM1583129,COSM3356083,COSM442676
S19	SNV	chr2	25457242	C	T	491	271	220	0.448	DNMT3A	ENSG00000119772	NON_SYNONYMOUS_CODING	ENST00000321117.5:c.2645G>A	ENSP00000324375.5:p.Arg882His	23/23	-	-	rs147001633,COSM9974O,COSM52944,COSM1583129,COSM3356083,COSM442676
S23	INDEL	chr2	25470488	A	-T	625	317	308	0.493	DNMT3A	ENSG00000119772	FRAME_SHIFT	ENST00000264709.3:c.986delT	ENSP00000264709.3:p.Met329SerfsTer16	8/23	YES	-	COSM1725001,COSM1725002
S23	INDEL	chr2	25470488	A	-T	625	317	308	0.493	DNMT3A	ENSG00000119772	FRAME_SHIFT	ENST00000380746.4:c.419delT	ENSP00000370122.4:p.Met140SerfsTer16	4/19	-	-	COSM1725001,COSM1725002
S23	INDEL	chr2	25470488	A	-T	625	317	308	0.493	DNMT3A	ENSG00000119772	FRAME_SHIFT	ENST00000321117.5:c.986delT	ENSP00000324375.5:p.Met329SerfsTer16	8/23	-	-	COSM1725001,COSM1725002
S23	INDEL	chr2	25470488	A	-T	625	317	308	0.493	DNMT3A	ENSG00000119772	FRAME_SHIFT	ENST00000402667.1:c.317delT	ENSP00000384237.1:p.Met106SerfsTer16	3/18	-	-	COSM1725001,COSM1725002
S25	SNV	chr2	25457242	C	G	233	153	80	0.343	DNMT3A	ENSG00000119772	NON_SYNONYMOUS_CODING	ENST00000264709.3:c.2645G>C	ENSP00000264709.3:p.Arg882Pro	23/23	YES	-	rs147001633,COSM9974O,COSM52944,COSM1583129,COSM3356083,COSM442676
S25	SNV	chr2	25457242	C	G	233	153	80	0.343	DNMT3A	ENSG00000119772	NON_SYNONYMOUS_CODING	ENST00000402667.1:c.1976G>C	ENSP00000384237.1:p.Arg659Pro	18/18	-	-	rs147001633,COSM9974O,COSM52944,COSM1583129,COSM3356083,COSM442676
S25	SNV	chr2	25457242	C	G	233	153	80	0.343	DNMT3A	ENSG00000119772	NON_SYNONYMOUS_CODING	ENST00000380746.4:c.2078G>C	ENSP00000370122.4:p.Arg693Pro	19/19	-	-	rs147001633,COSM9974O,COSM52944,COSM1583129,COSM3356083,COSM442676
S25	SNV	chr2	25457242	C	G	233	153	80	0.343	DNMT3A	ENSG00000119772	NON_SYNONYMOUS_CODING	ENST00000321117.5:c.2645G>C	ENSP00000324375.5:p.Arg882Pro	23/23	-	-	rs147001633,COSM9974O,COSM52944,COSM1583129,COSM3356083,COSM442676
S29	SNV	chr2	25464439	G	C	270	145	125	0.463	DNMT3A	ENSG00000119772	NON_SYNONYMOUS_CODING	ENST00000380746.4:c.1507C>G	ENSP00000370122.4:p.Gln503Glu	13/19	-	-	-
S29	SNV	chr2	25464439	G	C	270	145	125	0.463	DNMT3A	ENSG00000119772	NON_SYNONYMOUS_CODING	ENST00000321117.5:c.2074C>G	ENSP00000324375.5:p.Gln692Glu	17/23	-	-	-
S29	SNV	chr2	25464439	G	C	270	145	125	0.463	DNMT3A	ENSG00000119772	NON_SYNONYMOUS_CODING	ENST00000264709.3:c.2074C>G	ENSP00000264709.3:p.Gln692Glu	17/23	YES	-	-
S29	SNV	chr2	25464439	G	C	270	145	125	0.463	DNMT3A	ENSG00000119772	NON_SYNONYMOUS_CODING	ENST00000402667.1:c.1405C>G	ENSP00000384237.1:p.Gln469Glu	12/18	-	-	-
S30	SNV	chr2	25463276	A	C	310	161	149	0.481	DNMT3A	ENSG00000119772	NON_SYNONYMOUS_CODING	ENST00000380746.4:c.1650T>G	ENSP00000370122.4:p.His550Gln	15/19	-	-	-
S30	SNV	chr2	25463276	A	C	310	161	149	0.481	DNMT3A	ENSG00000119772	NON_SYNONYMOUS_CODING	ENST00000264709.3:c.2217T>G	ENSP00000264709.3:p.His739Gln	19/23	YES	-	-
S30	SNV	chr2	25463276	A	C	310	161	149	0.481	DNMT3A	ENSG00000119772	NON_SYNONYMOUS_CODING	ENST00000321117.5:c.2217T>G	ENSP00000324375.5:p.His739Gln	19/23	-	-	-
S30	SNV	chr2	25463276	A	C	310	161	149	0.481	DNMT3A	ENSG00000119772	NON_SYNONYMOUS_CODING	ENST00000402667.1:c.1548T>G	ENSP00000384237.1:p.His516Gln	14/18	-	-	-
S32	INDEL	chr2	25463227	C	-AAAGAGCCAGAA	347	228	119	0.343	DNMT3A	ENSG00000119772	STOP_GAINED	ENST00000380746.4:c.1699delG	ENSP00000370122.4:p.Glu567ArgfsTer23	15/19	-	-	-
S32	INDEL	chr2	25463227	C	-AAAGAGCCAGAA	347	228	119	0.343	DNMT3A	ENSG00000119772	STOP_GAINED	ENST00000402667.1:c.1597delG	ENSP00000384237.1:p.Glu533ArgfsTer23	14/18	-	-	-
S32	INDEL	chr2	25463227	C	-AAAGAGCCAGAA	347	228	119	0.343	DNMT3A	ENSG00000119772	CODON_CHANGE_PLUS_CODON_DELETION	ENST00000380746.4:c.1699delG	ENSP00000370122.4:p.Glu567ArgfsTer23	15/19	-	-	-
S32	SNV	chr2	25463299	A	C	329	164	165	0.501	DNMT3A	ENSG00000119772	NON_SYNONYMOUS_CODING	ENST00000402667.1:c.1525T>G	ENSP00000384237.1:p.Phe509Val	14/18	-	-	rs149043640
S32	SNV	chr2	25463299	A	C	329	164	165	0.501	DNMT3A	ENSG00000119772	NON_SYNONYMOUS_CODING	ENST00000380746.4:c.1627T>G	ENSP00000370122.4:p.Phe543Val	15/19	-	-	rs149043640
S32	SNV	chr2	25463299	A	C	329	164	165	0.501	DNMT3A	ENSG00000119772	NON_SYNONYMOUS_CODING	ENST00000321117.5:c.2194T>G	ENSP00000324375.5:p.Phe732Val	19/23	-	-	rs149043640
S32	INDEL	chr2	25463227	C	-AAAGAGCCAGAA	347	228	119	0.343	DNMT3A	ENSG00000119772	CODON_CHANGE_PLUS_CODON_DELETION	ENST00000402667.1:c.1597delG	ENSP00000384237.1:p.Glu533ArgfsTer23	14/18	-	-	-
S32	INDEL	chr2	25463227	C	-AAAGAGCCAGAA	347	228	119	0.343	DNMT3A	ENSG00000119772	STOP_GAINED	ENST00000321117.5:c.2266delG	ENSP00000324375.5:p.Glu756ArgfsTer23	19/23	-	-	-
S32	INDEL	chr2	25463227	C	-AAAGAGCCAGAA	347	228	119	0.343	DNMT3A	ENSG00000119772	CODON_CHANGE_PLUS_CODON_DELETION	ENST00000264709.3:c.2266delG	ENSP00000264709.3:p.Glu756ArgfsTer23	19/23	YES	-	-
S32	INDEL	chr2	25463227	C	-AAAGAGCCAGAA	347	228	119	0.343	DNMT3A	ENSG00000119772	CODON_CHANGE_PLUS_CODON_DELETION	ENST00000321117.5:c.2266delG	ENSP00000324375.5:p.Glu756ArgfsTer23	19/23	-	-	-
S32	SNV	chr2	25463299	A	C	329	164	165	0.501	DNMT3A	ENSG00000119772	NON_SYNONYMOUS_CODING	ENST00000264709.3:c.2194T>G	ENSP00000264709.3:p.Phe732Val	19/23	YES	-	rs149043640
S32	INDEL	chr2	25463227	C	-AAAGAGCCAGAA	347	228	119	0.343	DNMT3A	ENSG00000119772	STOP_GAINED	ENST00000264709.3:c.2266delG	ENSP00000264709.3:p.Glu756ArgfsTer23	19/23	YES	-	-
S34	SNV	chr2	25457242	C	T	468	290	178	0.38	DNMT3A	ENSG00000119772	NON_SYNONYMOUS_CODING	ENST00000321117.5:c.2645G>A	ENSP00000324375.5:p.Arg882His	23/23	-	-	rs147001633,COSM9974O,COSM52944,COSM1583129,COSM3356083,COSM442676
S34	SNV	chr2	25457242	C	T	468	290	178	0.38	DNMT3A	ENSG00000119772	NON_SYNONYMOUS_CODING	ENST00000402667.1:c.1976G>A	ENSP00000384237.1:p.Arg659His	18/18	-	-	rs147001633,COSM9974O,COSM52944,COSM1583129,COSM3356083,COSM442676
S34	SNV	chr2	25457242	C	T	468	290	178	0.38	DNMT3A	ENSG00000119772	NON_SYNONYMOUS_CODING	ENST00000380746.4:c.2078G>A	ENSP00000370122.4:p.Arg693His	19/19	-	-	rs147001633,COSM9974O,COSM52944,COSM1583129,COSM3356083,COSM442676
S34	SNV	chr2	25457242	C	T	468	290	178	0.38	DNMT3A	ENSG00000119772	NON_SYNONYMOUS_CODING	ENST00000264709.3:c.2645G>A	ENSP00000264709.3:p.Arg882His	23/23	YES	-	rs147001633,COSM9974O,COSM52944,COSM1583129,COSM3356083,COSM442676
S36	SNV	chr2	25457243	G	A	559	283	276	0.494	DNMT3A	ENSG00000119772	NON_SYNONYMOUS_CODING	ENST00000380746.4:c.2077C>T	ENSP00000370122.4:p.Arg693Cys	19/19	-	-	rs377577594,COSM1583136,COSM87001,COSM53042,COSM1166704
S36	SNV	chr2	25457243	G	A	559	283	276	0.494	DNMT3A	ENSG00000119772	NON_SYNONYMOUS_CODING	ENST00000321117.5:c.2644C>T	ENSP00000324375.5:p.Arg882Cys	23/23	-	-	rs377577594,COSM1583136,COSM87001,COSM53042,COSM1166704
S36	SNV	chr2	25457243	G	A	559	283	276	0.494	DNMT3A	ENSG00000119772	NON_SYNONYMOUS_CODING	ENST00000264709.3:c.2644C>T	ENSP00000264709.3:p.Arg882Cys	23/23	YES	-	rs377577594,COSM1583136,COSM87001,COSM53042,COSM1166704
S36	SNV	chr2	25457243	G	A	559	283	276	0.494	DNMT3A	ENSG00000119772	NON_SYNONYMOUS_CODING	ENST00000402667.1:c.1975C>T	ENSP00000384237.1:p.Arg659Cys	18/18	-	-	rs377577594,COSM1583136,COSM87001,COSM53042,COSM1166704
S45	SNV	chr2	25463182	G	A	387	211	176	0.455	DNMT3A	ENSG00000119772	STOP_GAINED	ENST00000380746.4:c.1744C>T	ENSP00000370122.4:p.Arg582Ter	15/19	-	-	COSM231563,COSM1583105
S45	SNV	chr2	25463182	G	A	387	211	176	0.455	DNMT3A	ENSG00000119772	STOP_GAINED	ENST00000264709.3:c.2311C>T	ENSP00000264709.3:p.Arg771Ter	19/23	YES	-	COSM231563,COSM1583105
S45	SNV	chr2	25463182	G	A	387	211	176	0.455	DNMT3A	ENSG00000119772	STOP_GAINED	ENST00000402667.1:c.1642C>T	ENSP00000384237.1:p.Arg548Ter	14/18	-	-	COSM231563,COSM1583105
S45	SNV	chr2	25463182	G	A	387	211	176	0.455	DNMT3A	ENSG00000119772	STOP_GAINED	ENST00000321117.5:c.2311C>T	ENSP00000324375.5:p.Arg771Ter	19/23	-	-	COSM231563,COSM1583105
S46	SNV	chr2	25457242	C	T	419	193	226	0.539	DNMT3A	ENSG00000119772	NON_SYNONYMOUS_CODING	ENST00000380746.4:c.2078G>A	ENSP00000370122.4:p.Arg693His	19/19	-	-	rs147001633,COSM9974O,COSM52944,COSM1583129,COSM3356083,COSM442676
S46	SNV	chr2	25457242	C	T	419	193	226	0.539	DNMT3A	ENSG00000119772	NON_SYNONYMOUS_CODING	ENST00000264709.3:c.2645G>A	ENSP00000264709.3:p.Arg882His	23/23	YES	-	rs147001633,COSM9974O,COSM52944,COSM1583129,COSM3356083,COSM442676
S46	SNV	chr2	25457242	C	T	419	193	226	0.539	DNMT3A	ENSG00000119772	NON_SYNONYMOUS_CODING	ENST00000402667.1:c.1976G>A	ENSP00000384237.1:p.Arg659His	18/18	-	-	rs147001633,COSM9974O,COSM52944,COSM1583129,COSM3356083,COSM442676
S46	SNV	chr2	25457242	C	T	419	193	226	0.539	DNMT3A	ENSG00000119772	NON_SYNONYMOUS_CODING	ENST00000321117.5:c.2645G>A	ENSP00000324375.5:p.Arg882His	23/23	-	-	rs147001633,COSM9974O,COSM52944,COSM1583129,COSM3356083,COSM442676
S47	SNV	chr2	25470497	C	T	367	193	174	0.474	DNMT3A	ENSG00000119772	NON_SYNONYMOUS_CODING	ENST00000380746.4:c.410G>A	ENSP00000370122.4:p.Arg137His	4/19	-	-	COSM477214
S47	SNV	chr2	25470497	C	T	367	193	174	0.474	DNMT3A	ENSG00000119772	NON_SYNONYMOUS_CODING	ENST00000321117.5:c.977G>A	ENSP00000324375.5:p.Arg326His	8/23	-	-	COSM477214
S47	SNV	chr2	25470497	C	T	367	193	174	0.474	DNMT3A	ENSG00000119772	NON_SYNONYMOUS_CODING	ENST00000402667.1:c.308G>A	ENSP00000384237.1:p.Arg103His	3/18	-	-	COSM477214
S47	SNV	chr2	25470497	C	T	367	193	174	0.474	DNMT3A	ENSG00000119772	NON_SYNONYMOUS_CODING	ENST00000264709.3:c.977G>A	ENSP00000264709.3:p.Arg326His	8/23	YES	-	COSM477214
S53	SNV	chr2	25457242	C	T	470	251	219	0.466	DNMT3A	ENSG00000119772	NON_SYNONYMOUS_CODING	ENST00000321117.5:c.2645G>A	ENSP00000324375.5:p.Arg882His	23/23	-	-	rs147001633,COSM9974O,COSM52944,COSM1583129,COSM3356083,COSM442676
S53	SNV	chr2	25457242	C	T	470	251	219	0.466	DNMT3A	ENSG00000119772	NON_SYNONYMOUS_CODING	ENST00000380746.4:c.2078G>A	ENSP00000370122.4:p.Arg693His	19/19	-	-	rs147001633,COSM9974O,COSM52944,COSM1583129,COSM3356083,COSM442676
S53	SNV	chr2	25457242	C	T	470	251	219	0.466	DNMT3A	ENSG00000119772	NON_SYNONYMOUS_CODING	ENST00000264709.3:c.2645G>A	ENSP00000264709.3:p.Arg882His	23/23	YES	-	rs147001633,COSM9974O,COSM52944,COSM1583129,COSM3356083,COSM442676
S53	SNV	chr2	25457242	C	T	470	251	219	0.466	DNMT3A	ENSG00000119772	NON_SYNONYMOUS_CODING	ENST00000402667.1:c.1976G>A	ENSP00000384237.1:p.Arg659His	18/18	-	-	rs147001633,COSM9974O,COSM52944,COSM1583129,COSM3356083,COSM442676
S57	SNV	chr2	25457242	C	T	674	364	310	0.46	DNMT3A	ENSG00000119772	NON_SYNONYMOUS_CODING	ENST00000321117.5:c.2645G>A	ENSP00000324375.5:p.Arg882His	23/23	-	-	rs147001633,COSM9974O,COSM52944,COSM1583129,COSM3356083,COSM442676
S57	SNV	chr2	25457242	C	T	674	364	310	0.46	DNMT3A	ENSG00000119772	NON_SYNONYMOUS_CODING	ENST00000264709.3:c.2645G>A	ENSP00000264709.3:p.Arg882His	23/23	YES	-	rs147001633,COSM9974O,COSM52944,COSM1583129,COSM3356083,COSM442676
S57	SNV	chr2	25457242	C	T	674	364	310	0.46	DNMT3A	ENSG00000119772	NON_SYNONYMOUS_CODING	ENST00000380746.4:c.2078G>A	ENSP00000370122.4:p.Arg693His	19/19	-	-	rs147001633,COSM9974O,COSM52944,COSM1583129,COSM3356083,COSM442676
S57	SNV	chr2	25457242	C	T	674	364	310	0.46	DNMT3A	ENSG00000119772	NON_SYNONYMOUS_CODING	ENST00000402667.1:c.1976G>A	ENSP00000384237.1:p.Arg659His	18/18	-	-	rs147001633,COSM9974O,COSM52944,COSM1583129,COSM3356083,COSM442676
S58	SNV	chr2	25457242	C	T	727	355	372	0.512	DNMT3A	ENSG00000119772	NON_SYNONYMOUS_CODING	ENST00000321117.5:c.2645G>A	ENSP00000324375.5:p.Arg882His	23/23	-	-	rs147001633,COSM9974O,COSM52944,COSM1583129,COSM3356083,COSM442676
S58	SNV	chr2	25457242	C	T	727	355	372	0.512	DNMT3A	ENSG00000119772	NON_SYNONYMOUS_CODING	ENST00000380746.4:c.2078G>A	ENSP00000370122.4:p.Arg693His	19/19	-	-	rs147001633,COSM9974O,COSM52944,COSM1583129,COSM3356083,COSM442676
S58	SNV	chr2	25457242	C	T	727	355	372	0.512	DNMT3A	ENSG00000119772	NON_SYNONYMOUS_CODING	ENST00000402667.1:c.1976G>A	ENSP00000384237.1:p.Arg659His	18/18	-	-	rs147001633,COSM9974O,COSM52944,COSM1583129,COSM3356083,COSM442676
S58	SNV	chr2	25457242	C	T	727	355	372	0.512	DNMT3A	ENSG00000119772	NON_SYNONYMOUS_CODING	ENST00000264709.3:c.2645G>A	ENSP00000264709.3:p.Arg882His	23/23	YES	-	rs147001633,COSM9974O,COSM52944,COSM1583129,COSM3356083,COSM442676
S63	SNV	chr2	25466834	G	C	340	231	109	0.321	DNMT3A	ENSG00000119772	STOP_GAINED	ENST00000264709.3:c.1869C>G	ENSP00000264709.3:p.Tyr623Ter	16/23	YES	-	COSM144489
S63	SNV	chr2	25466834	G	C	340	231	109	0.321	DNMT3A	ENSG00000119772	STOP_GAINED	ENST00000402667.1:c.1200C>G	ENSP00000384237.1:p.Tyr400Ter	11/18	-	-	COSM144489
S63	SNV	chr2	25466834	G	C	340	231	109	0.321	DNMT3A	ENSG00000119772	STOP_GAINED	ENST00000321117.5:c.1869C>G	ENSP00000324375.5:p.Tyr623Ter	16/23	-	-	COSM144489
S63	SNV	chr2	25466834	G	C	340	231	109	0.321	DNMT3A	ENSG00000119772	STOP_GAINED	ENST00000380746.4:c.1302C>G	ENSP00000370122.4:p.Tyr434Ter	12/19	-	-	COSM144489
S65	SNV	chr2	25457243	G	A	582	322	260	0.447	DNMT3A	ENSG00000119772	NON_SYNONYMOUS_CODING	ENST00000402667.1:c.1975C>T	ENSP00000384237.1:p.Arg659Cys	18/18	-	-	rs377577594,COSM1583136,COSM87001,COSM53042,COSM1166704
S65	SNV	chr2	25457243	G	A	582	322	260	0.447	DNMT3A	ENSG00000119772	NON_SYNONYMOUS_CODING	ENST00000380746.4:c.2077C>T	ENSP00000370122.4:p.Arg693Cys	19/19	-	-	rs377577594,COSM1583136,COSM87001,COSM53042,COSM1166704
S65	SNV	chr2	25457243	G	A	582	322	260	0.447	DNMT3A	ENSG00000119772	NON_SYNONYMOUS_CODING	ENST00000321117.5:c.2644C>T	ENSP00000324375.5:p.Arg882Cys	23/23	-	-	rs377577594,COSM1583136,COSM87001,COSM53042,COSM1166704
S65	SNV	chr2	25457243	G	A	582	322	260	0.447	DNMT3A	ENSG00000119772	NON_SYNONYMOUS_CODING	ENST00000264709.3:c.2644C>T	ENSP00000264709.3:p.Arg882Cys	23/23	YES	-	rs377577594,COSM1583136,COSM87001,COSM53042,COSM1166704
1	SNV	chr20	32264462	C	T	123	74	49	0.398	E2F1	ENSG00000101412	UTR_3_PRIME	ENST00000343380.5:c.*76G>A	-	7/7	YES	0.0002	rs532185356
64	SNV	chr20	32266124	A	G	221	123	98	0.443	E2F1	ENSG00000101412	NON_SYNONYMOUS_CODING	ENST00000343380.5:c.608T>C	ENSP00000345571.5:p.Leu203Pro	4/7	YES	0.0002	rs546049774
S22	INDEL	chr20	32266127	C	-G	438	290	148	0.337	E2F1	ENSG00000101412	FRAME_SHIFT	ENST00000343380.5:c.606delG	ENSP00000345571.5:p.Glu204ArgfsTer3	4/7	YES	-	rs377637237
S36	SNV	chr5	158250225	C	T	867	468	399	0.46	EBF1	ENSG00000164330	NON_SYNONYMOUS_CODING	ENST00000513708.6:c.737G>A	ENSP00000322898.6:p.Arg246Lys	8/16	YES	-	-
S36	SNV	chr5	158250225	C	T	867	468	399	0.46	EBF1	ENSG00000164330	NON_SYNONYMOUS_CODING	ENST00000509054.4:c.668G>A	ENSP00000370029.4:p.Arg223Lys	7/15	-	-	-
S36	SNV	chr5	158250225	C	T	867	468	399	0.46	EBF1	ENSG00000164330	NON_SYNONYMOUS_CODING	ENST00000517373.1:c.737G>A	ENSP00000428020.1:p.Arg246Lys	8/15	-	-	-
8	SNV	chr6	139170538	G	A	821	429	392	0.477	ECT2L	ENSG00000203734	NON_SYNONYMOUS_CODING	ENST00000423192.1:c.1036G>A	ENSP00000387368.1:p.Asp346Asn	8/21	YES	-	-
8	SNV	chr6	139170538	G	A	821	429	392	0.477	ECT2L	ENSG00000203734	NON_SYNONYMOUS_CODING	ENST00000541398.1:c.829G>A	ENSP00000442307.1:p.Asp277Asn	6/17	-	-	-
8	SNV	chr6	139170538	G	A	821	429	392	0.477	ECT2L	ENSG00000203734	NON_SYNONYMOUS_CODING	ENST00000367682.2:c.1036G>A	ENSP00000356655.2:p.Asp346Asn	7/20	-	-	-
S20	SNV	chr6	139165601	C	G	965	452	513	0.532	ECT2L	ENSG00000203734	NON_SYNONYMOUS_CODING	ENST00000367682.2:c.648C>G	ENSP00000356655.2:p.Ser216Arg	5/20	-	0.0002	rs549245814
S20	SNV	chr6	139165601	C	G	965	452	513	0.532	ECT2L	ENSG00000203734	NON_SYNONYMOUS_CODING	ENST00000541398.1:c.441C>G	ENSP00000442307.1:p.Ser147Arg	4/17	-	0.0002	rs549245814
S20	SNV	chr6	139165601	C	G	965	452	513	0.532	ECT2L	ENSG00000203734	NON_SYNONYMOUS_CODING	ENST00000423192.1:c.648C>G	ENSP00000387368.1:p.Ser216Arg	6/21	YES	0.0002	rs549245814
7	SNV	chr19	18555667	G	C	212	127	85	0.401	ELL	ENSG00000105656	NON_SYNONYMOUS_CODING	ENST00000262809.4:c.1751G>C	ENSP00000262809.3:p.Arg584Pro	12/12	YES	0.0006	rs568878298
S14	SNV	chr19	18572617	G	A	178	84	94	0.528	ELL	ENSG00000105656	NON_SYNONYMOUS_CODING	ENST00000596124.3:c.116C>T	ENSP00000475648.2:p.Ala39Val	5/12	-	-	-
S14	SNV	chr19	18572617	G	A	178	84	94	0.528	ELL	ENSG00000105656	NON_SYNONYMOUS_CODING	ENST00000262809.4:c.515C>T	ENSP00000262809.3:p.Ala172Val	5/12	YES	-	-
S59	SNV	chr19	18561673	T	C	590	284	306	0.518	ELL	ENSG00000105656	NON_SYNONYMOUS_CODING	ENST00000262809.4:c.1079A>G	ENSP00000262809.3:p.Asn360Ser	8/12	YES	0.0002	rs553696102
S59	SNV	chr19	18561673	T	C	590	284	306	0.518	ELL	ENSG00000105656	NON_SYNONYMOUS_CODING	ENST00000596124.3:c.680A>G	ENSP00000475648.2:p.Asn227Ser	8/12	-	0.0002	rs553696102
64	SNV	chr7	73470748	C	T	137	61	76	0.555	ELN	ENSG00000049540	NON_SYNONYMOUS_CODING	ENST00000380553.4:c.947C>T	ENSP00000369926.4:p.Pro316Leu	14/25	-	-	-
64	SNV	chr7	73470748	C	T	137	61	76	0.555	ELN	ENSG00000049540	NON_SYNONYMOUS_CODING	ENST00000357036.5:c.1313C>T	ENSP00000349540.5:p.Pro438Leu	20/32	-	-	-
64	SNV	chr7	73470748	C	T	137	61	76	0.555	ELN	ENSG00000049540	NON_SYNONYMOUS_CODING	ENST00000414324.1:c.1283C>T	ENSP00000392575.1:p.Pro428Leu	19/31	-	-	-
64	SNV	chr7	73470748	C	T	137	61	76	0.555	ELN	ENSG00000049540	NON_SYNONYMOUS_CODING	ENST00000458204.1:c.1268C>T	ENSP00000403162.1:p.Pro423Leu	19/32	-	-	-

64	SNV	chr7	73470748	C	T	137	61	76	0.555	ELN	ENSG00000049540	NON_SYNONYMOUS_CODING	ENST00000320399.6:c.1298C>T	ENSP00000311566.6:p.Pro433Leu	20/33	-	-	-
64	SNV	chr7	73470748	C	T	137	61	76	0.555	ELN	ENSG00000049540	NON_SYNONYMOUS_CODING	ENST00000429192.1:c.1313C>T	ENSP00000391129.1:p.Pro438Leu	20/31	-	-	-
64	SNV	chr7	73470748	C	T	137	61	76	0.555	ELN	ENSG00000049540	NON_SYNONYMOUS_CODING	ENST00000380575.4:c.1298C>T	ENSP00000369949.4:p.Pro423Leu	19/30	-	-	-
64	SNV	chr7	73470748	C	T	137	61	76	0.555	ELN	ENSG00000049540	NON_SYNONYMOUS_CODING	ENST00000380576.5:c.1298C>T	ENSP00000369950.5:p.Pro433Leu	20/32	-	-	-
64	SNV	chr7	73470748	C	T	137	61	76	0.555	ELN	ENSG00000049540	NON_SYNONYMOUS_CODING	ENST00000445912.1:c.1298C>T	ENSP00000389857.1:p.Pro433Leu	20/32	-	-	-
64	SNV	chr7	73470748	C	T	137	61	76	0.555	ELN	ENSG00000049540	NON_SYNONYMOUS_CODING	ENST00000358929.4:c.1298C>T	ENSP00000351807.4:p.Pro433Leu	20/34	-	-	-
64	SNV	chr7	73470748	C	T	137	61	76	0.555	ELN	ENSG00000049540	NON_SYNONYMOUS_CODING	ENST00000252034.7:c.1298C>T	ENSP00000252034.7:p.Pro433Leu	20/33	YES	-	-
64	SNV	chr7	73470748	C	T	137	61	76	0.555	ELN	ENSG00000049540	NON_SYNONYMOUS_CODING	ENST00000380584.4:c.1256C>T	ENSP00000369956.4:p.Pro419Leu	19/29	-	-	-
64	SNV	chr7	73470748	C	T	137	61	76	0.555	ELN	ENSG00000049540	NON_SYNONYMOUS_CODING	ENST00000380562.4:c.1298C>T	ENSP00000369936.4:p.Pro433Leu	20/33	-	-	-
S54	SNV	chr7	73477690	G	A	319	161	158	0.495	ELN	ENSG00000049540	NON_SYNONYMOUS_CODING	ENST00000380584.4:c.1785G>A	ENSP00000369958.4:p.Ala589Thr	25/29	-	-	-
S54	SNV	chr7	73477690	G	A	319	161	158	0.495	ELN	ENSG00000049540	NON_SYNONYMOUS_CODING	ENST00000320492.7:c.1666G>A	ENSP00000315607.7:p.Ala556Thr	24/29	-	-	-
S54	SNV	chr7	73477690	G	A	319	161	158	0.495	ELN	ENSG00000049540	NON_SYNONYMOUS_CODING	ENST00000414324.1:c.1837G>A	ENSP00000392575.1:p.Ala613Thr	26/31	-	-	-
S54	SNV	chr7	73477690	G	A	319	161	158	0.495	ELN	ENSG00000049540	NON_SYNONYMOUS_CODING	ENST00000380562.4:c.1927G>A	ENSP00000369936.4:p.Ala643Thr	28/33	-	-	-
S54	SNV	chr7	73477690	G	A	319	161	158	0.495	ELN	ENSG00000049540	NON_SYNONYMOUS_CODING	ENST00000358929.4:c.2113G>A	ENSP00000351807.4:p.Ala705Thr	29/34	-	-	-
S54	SNV	chr7	73477690	G	A	319	161	158	0.495	ELN	ENSG00000049540	NON_SYNONYMOUS_CODING	ENST00000458204.1:c.1879G>A	ENSP00000403162.1:p.Ala627Thr	27/32	-	-	-
S54	SNV	chr7	73477690	G	A	319	161	158	0.495	ELN	ENSG00000049540	NON_SYNONYMOUS_CODING	ENST00000320399.6:c.2008G>A	ENSP00000313565.6:p.Ala670Thr	28/33	-	-	-
S54	SNV	chr7	73477690	G	A	319	161	158	0.495	ELN	ENSG00000049540	NON_SYNONYMOUS_CODING	ENST00000429192.1:c.1867G>A	ENSP00000391129.1:p.Ala623Thr	27/31	-	-	-
S54	SNV	chr7	73477690	G	A	319	161	158	0.495	ELN	ENSG00000049540	NON_SYNONYMOUS_CODING	ENST00000252034.7:c.1909G>A	ENSP00000252034.7:p.Ala637Thr	28/33	YES	-	-
S54	SNV	chr7	73477690	G	A	319	161	158	0.495	ELN	ENSG00000049540	NON_SYNONYMOUS_CODING	ENST00000380576.5:c.1852G>A	ENSP00000369950.5:p.Ala618Thr	27/32	-	-	-
S54	SNV	chr7	73477690	G	A	319	161	158	0.495	ELN	ENSG00000049540	NON_SYNONYMOUS_CODING	ENST00000380553.4:c.1501G>A	ENSP00000369926.4:p.Ala501Thr	21/25	-	-	-
S54	SNV	chr7	73477690	G	A	319	161	158	0.495	ELN	ENSG00000049540	NON_SYNONYMOUS_CODING	ENST00000380575.4:c.1822G>A	ENSP00000369949.4:p.Ala608Thr	26/30	-	-	-
S54	SNV	chr7	73477690	G	A	319	161	158	0.495	ELN	ENSG00000049540	NON_SYNONYMOUS_CODING	ENST00000445912.1:c.1909G>A	ENSP00000389857.1:p.Ala637Thr	28/32	-	-	-
S54	SNV	chr7	73477690	G	A	319	161	158	0.495	ELN	ENSG00000049540	NON_SYNONYMOUS_CODING	ENST00000357036.5:c.1924G>A	ENSP00000349540.5:p.Ala642Thr	28/32	-	-	-
S22	SNV	chr2	42522551	T	G	655	331	324	0.495	EML4	ENSG00000143924	NON_SYNONYMOUS_CODING	ENST00000318522.5:c.1384T>G	ENSP00000320663.5:p.Leu462Val	13/23	YES	-	-
S22	SNV	chr2	42522551	T	G	655	331	324	0.495	EML4	ENSG00000143924	NON_SYNONYMOUS_CODING	ENST00000402711.2:c.1210T>G	ENSP00000385059.2:p.Leu404Val	12/22	-	-	-
S22	SNV	chr2	42522551	T	G	655	331	324	0.495	EML4	ENSG00000143924	NON_SYNONYMOUS_CODING	ENST00000401738.3:c.1417T>G	ENSP00000384939.3:p.Leu473Val	14/24	-	-	-
19	INDEL	chr22	41574340	A	-ACCAGTTCCAGC	814	515	299	0.367	EP300	ENSG00000100393	CODON_DELETION	ENST00000263253.7:c.6626delA	ENSP00000263253.7:p.Asn2209ThrfsTer34	31/31	YES	-	-
25	INDEL	chr22	41574281	T	-GCAACAGCAGCA	603	402	201	0.333	EP300	ENSG00000100393	CODON_CHANGE_PLUS_CODON_DELETION	ENST00000263253.7:c.6567delG	ENSP00000263253.7:p.Met2189IlefsTer13	31/31	YES	-	-
30	SNV	chr22	41536257	A	G	196	125	71	0.362	EP300	ENSG00000100393	NON_SYNONYMOUS_CODING	ENST00000263253.7:c.1874A>G	ENSP00000263253.7:p.Asn625Ser	9/31	YES	-	-
8	INDEL	chr22	41574340	A	-ACCAGTTCCAGC	1531	1172	359	0.234	EP300	ENSG00000100393	CODON_DELETION	ENST00000263253.7:c.6626delA	ENSP00000263253.7:p.Asn2209ThrfsTer34	31/31	YES	-	-
S54	INDEL	chr22	41574340	A	-ACCAGTTCCAGC	707	511	196	0.277	EP300	ENSG00000100393	CODON_DELETION	ENST00000263253.7:c.6626delA	ENSP00000263253.7:p.Asn2209ThrfsTer34	31/31	YES	-	-
S56	SNV	chr22	41564741	G	T	1056	610	446	0.422	EP300	ENSG00000100393	STOP_GAINED	ENST00000263253.7:c.4042G>T	ENSP00000263253.7:p.Glu1348Ter	25/31	YES	-	-
46	SNV	chr19	11491756	G	T	131	65	66	0.504	EPOR	ENSG00000187266	NON_SYNONYMOUS_CODING	ENST00000222139.6:c.715C>G	ENSP00000222139.5:p.Pro239Ala	8/8	YES	-	-
S52	SNV	chr19	11489237	C	A	470	259	211	0.449	EPOR	ENSG00000187266	NON_SYNONYMOUS_CODING	ENST00000222139.6:c.950G>T	ENSP00000222139.5:p.Trp317Leu	8/8	YES	-	-
S58	SNV	chr19	11488727	T	C	737	400	337	0.457	EPOR	ENSG00000187266	NON_SYNONYMOUS_CODING	ENST00000222139.6:c.1460A>G	ENSP00000222139.5:p.Asn487Ser	8/8	YES	0.0022	rs62638745,CM962728
23	SNV	chr17	37868656	A	G	235	121	114	0.485	ERBB2	ENSG00000141736	NON_SYNONYMOUS_CODING	ENST00000541774.1:c.1058A>G	ENSP00000446466.1:p.Lys353Arg	9/27	-	-	-
23	SNV	chr17	37868656	A	G	235	121	114	0.485	ERBB2	ENSG00000141736	NON_SYNONYMOUS_CODING	ENST00000445658.2:c.275A>G	ENSP00000404047.2:p.Lys92Arg	3/21	-	-	-
23	SNV	chr17	37868656	A	G	235	121	114	0.485	ERBB2	ENSG00000141736	NON_SYNONYMOUS_CODING	ENST00000406381.2:c.1013A>G	ENSP00000385185.2:p.Lys338Arg	11/29	-	-	-
23	SNV	chr17	37868656	A	G	235	121	114	0.485	ERBB2	ENSG00000141736	NON_SYNONYMOUS_CODING	ENST00000540147.1:c.1013A>G	ENSP00000443562.1:p.Lys338Arg	9/27	-	-	-
23	SNV	chr17	37868656	A	G	235	121	114	0.485	ERBB2	ENSG00000141736	NON_SYNONYMOUS_CODING	ENST00000269571.5:c.1103A>G	ENSP00000269571.4:p.Lys368Arg	9/27	YES	-	-
23	SNV	chr17	37868656	A	G	235	121	114	0.485	ERBB2	ENSG00000141736	NON_SYNONYMOUS_CODING	ENST00000540042.1:c.1013A>G	ENSP00000446382.1:p.Lys338Arg	9/15	-	-	-
43	SNV	chr17	37866402	G	A	381	186	195	0.512	ERBB2	ENSG00000141736	NON_SYNONYMOUS_CODING	ENST00000406381.2:c.617G>A	ENSP00000385185.2:p.Cys206Tyr	8/29	-	-	-
43	SNV	chr17	37866402	G	A	381	186	195	0.512	ERBB2	ENSG00000141736	NON_SYNONYMOUS_CODING	ENST00000540042.1:c.617G>A	ENSP00000446382.1:p.Cys206Tyr	6/15	-	-	-
43	SNV	chr17	37866402	G	A	381	186	195	0.512	ERBB2	ENSG00000141736	NON_SYNONYMOUS_CODING	ENST00000269571.5:c.707G>A	ENSP00000269571.4:p.Cys236Tyr	6/27	YES	-	-
43	SNV	chr17	37866402	G	A	381	186	195	0.512	ERBB2	ENSG00000141736	NON_SYNONYMOUS_CODING	ENST00000540147.1:c.617G>A	ENSP00000443562.1:p.Cys206Tyr	6/27	-	-	-
43	SNV	chr17	37866402	G	A	381	186	195	0.512	ERBB2	ENSG00000141736	NON_SYNONYMOUS_CODING	ENST00000541774.1:c.662G>A	ENSP00000446466.1:p.Cys221Tyr	6/27	-	-	-
63	SNV	chr17	37882045	G	C	454	236	218	0.48	ERBB2	ENSG00000141736	NON_SYNONYMOUS_CODING	ENST00000269571.5:c.2811G>C	ENSP00000269571.4:p.Lys937Asn	23/27	YES	-	COSM3516815
63	SNV	chr17	37882045	G	C	454	236	218	0.48	ERBB2	ENSG00000141736	NON_SYNONYMOUS_CODING	ENST00000406381.2:c.2721G>C	ENSP00000385185.2:p.Lys907Asn	25/29	-	-	COSM3516815
63	SNV	chr17	37882045	G	C	454	236	218	0.48	ERBB2	ENSG00000141736	NON_SYNONYMOUS_CODING	ENST00000445658.2:c.1983G>C	ENSP00000404047.2:p.Lys661Asn	17/21	-	-	COSM3516815
63	SNV	chr17	37882045	G	C	454	236	218	0.48	ERBB2	ENSG00000141736	NON_SYNONYMOUS_CODING	ENST00000540147.1:c.2721G>C	ENSP00000443562.1:p.Lys907Asn	23/27	-	-	COSM3516815
63	SNV	chr17	37882045	G	C	454	236	218	0.48	ERBB2	ENSG00000141736	NON_SYNONYMOUS_CODING	ENST00000541774.1:c.2766G>C	ENSP00000446466.1:p.Lys922Asn	23/27	-	-	COSM3516815
1	SNV	chr19	45860581	C	T	346	182	164	0.474	ERCC2	ENSG00000104884	NON_SYNONYMOUS_CODING	ENST00000391941.2:c.1354G>A	ENSP00000375805.2:p.Val452Ile	14/21	-	0.0018	rs531021258
1	SNV	chr19	45860581	C	T	346	182	164	0.474	ERCC2	ENSG00000104884	NON_SYNONYMOUS_CODING	ENST00000391944.3:c.1192G>A	ENSP00000375808.3:p.Val398Ile	13/21	-	0.0018	rs531021258
1	SNV	chr19	45860581	C	T	346	182	164	0.474	ERCC2	ENSG00000104884	NON_SYNONYMOUS_CODING	ENST00000391945.4:c.1426G>A	ENSP00000375809.3:p.Val476Ile	15/23	YES	0.0018	rs531021258
38	SNV	chr19	45855826	G	A	664	354	310	0.467	ERCC2	ENSG00000104884	STOP_GAINED	ENST00000391944.3:c.1750C>T	ENSP00000375808.3:p.Gln584Ter	19/21	-	-	CM091461
38	SNV	chr19	45855826	G	A	664	354	310	0.467	ERCC2	ENSG00000104884	STOP_GAINED	ENST00000391941.2:c.1912C>T	ENSP00000375805.2:p.Gln638Ter	20/21	-	-	CM091461
38	SNV	chr19	45855826	G	A	664	354	310	0.467	ERCC2	ENSG00000104884	STOP_GAINED	ENST00000391945.4:c.1984C>T	ENSP00000375809.3:p.Gln662Ter	21/23	YES	-	CM091461
40	SNV	chr19	45864820	A	C	159	92	67	0.421	ERCC2	ENSG00000104884	NON_SYNONYMOUS_CODING	ENST00000391941.2:c.1127T>G	ENSP00000375805.2:p.Leu376Arg	11/21	-	-	-
40	SNV	chr19	45864820	A	C	159	92	67	0.421	ERCC2	ENSG00000104884	NON_SYNONYMOUS_CODING	ENST00000391944.3:c.965T>G	ENSP00000375808.3:p.Leu322Arg	10/21	-	-	-
40	SNV	chr19	45864820	A	C	159	92	67	0.421	ERCC2	ENSG00000104884	NON_SYNONYMOUS_CODING	ENST00000391945.4:c.1199T>G	ENSP00000375809.3:p.Leu400Arg	12/23	YES	-	-
40	SNV	chr19	45864820	A	C	159	92	67	0.421	ERCC2	ENSG00000104884	NON_SYNONYMOUS_CODING	ENST00000485403.2:c.1127T>G	ENSP00000431229.2:p.Leu376Arg	11/12	-	-	-
40	SNV	chr19	45864820	A	C	159	92	67	0.421	ERCC2	ENSG00000104884	NON_SYNONYMOUS_CODING	ENST00000391940.4:c.1127T>G	ENSP00000375804.4:p.Leu376Arg	12/13	-	-	-
S10	INDEL	chr19	45856553	T	-AA	216	106	110	0.509	ERCC2	ENSG00000104884	FRAME_SHIFT	ENST00000391945.4:c.1705delA	ENSP00000375809.3:p.Ile569LeufsTer140	18/23	YES	0.0002	rs141944141,CD013475
S10	INDEL	chr19	45856553	T	-AA	216	106	110	0.509	ERCC2	ENSG00000104884	FRAME_SHIFT	ENST00000391941.2:c.1633delA	ENSP00000375805.2:p.Ile545LeufsTer140	17/21	-	0.0002	rs141944141,CD013475
S10	INDEL	chr19	45856553	T	-AA	216	106	110	0.509	ERCC2	ENSG00000104884	FRAME_SHIFT	ENST00000391944.3:c.1471delA	ENSP00000375808.3:p.Ile491LeufsTer140	16/21	-	0.0002	rs141944141,CD013475
S20	SNV	chr19	45871950	C	T	959	488	471	0.491	ERCC2	ENSG00000104884	NON_SYNONYMOUS_CODING	ENST00000586131.1:c.226G>A	ENSP00000464887.1:p.Glu76Lys	5/10	-	-	-
S20	SNV	chr19	45871950	C	T	959	488	471	0.491	ERCC2	ENSG00000104884	NON_SYNONYMOUS_CODING	ENST00000391940.4:c.226G>A	ENSP00000375804.4:p.Glu76Lys	5/13	-	-	-
S20	SNV	chr19	45871950	C	T	959	488	471	0.491	ERCC2	ENSG00000104884	NON_SYNONYMOUS_CODING	ENST00000485403.2:c.226G>A	ENSP00000431229.2:p.Glu76Lys	4/12	-	-	-
S20	SNV	chr19	45871950	C	T	959	488	471	0.491	ERCC2	ENSG00000104884	NON_SYNONYMOUS_CODING	ENST00000391945.4:c.298G>A	ENSP00000375809.3:p.Glu100Lys	5/23	YES	-	-
S20	SNV	chr19	45871950	C	T	959	488	471	0.491	ERCC2	ENSG00000104884	NON_SYNONYMOUS_CODING	ENST00000586856.1:c.148G>A	ENSP00000466998.1:p.Glu50Lys	3/4	-	-	-
S20	SNV	chr19	45871950	C	T	959	488	471	0.491	ERCC2	ENSG00000104884	NON_SYNONYMOUS_CODING	ENST00000391941.2:c.226G>A	ENSP00000375805.2:p.Glu76Lys	4/21	-	-	-
S20	SNV	chr19	45871950	C	T	959	488	471	0.491	ERCC2	ENSG00000104884	NON_SYNONYMOUS_CODING	ENST00000221481.6:c.298G>A	ENSP00000221481.6:p.Glu100Lys	5/8	-	-	-
S20	SNV	chr19	45871950	C	T	959	488	471	0.491	ERCC2	ENSG00000104884	NON_SYNONYMOUS_CODING	ENST00000391944.3:c.298G>A	ENSP00000375808.3:p.Glu100Lys	5/21	-	-	-
S22	SNV	chr19	45856577	T	C	296	160	136	0.46	ERCC2	ENSG00000104884	NON_SYNONYMOUS_CODING	ENST00000391944.3:c.1447A>G	ENSP00000375808.3:p.Ile483Val	16/21	-	-	-
S22	SNV	chr19	45856577	T	C	296	160	136	0.46	ERCC2	ENSG00000104884	NON_SYNONYMOUS_CODING	ENST00000391945.4:c.1681A>G	ENSP00000375809.3:p.Ile561Val	18/23	YES	-	-
S22	SNV	chr19	45856577	T	C	296	160	136	0.46	ERCC2	ENSG00000104884	NON_SYNONYMOUS_CODING	ENST00000391941.2:c.1609A>G	ENSP00000375805.2:p.Ile537Val	17/21	-	-	-
S39	SNV	chr19	45867535	C	T	301	159	142	0.472	ERCC2	ENSG00000104884	NON_SYNONYMOUS_CODING	ENST00000485403.2:c.701G>A	ENSP00000431229.2:p.Arg234Gln	8/12	-	-	-
S39	SNV	chr19	45867535	C	T	301	159	142	0.472	ERCC2	ENSG00000104884	NON_SYNONYMOUS_CODING	ENST00000391944.3:c.539G>A	ENSP00000375808.3:p.Arg180Gln	7/21	-	-	-
S39	SNV	chr19	45867535	C	T	301	159	142	0.472	ERCC2	ENSG00000104884	NON_SYNONYMOUS_CODING	ENST00000391940.4:c.701G>A	ENSP00000375804.4:p.Arg234Gln	9/13	-	-	-
S39	SNV	chr19	45867535	C	T	301	159	142	0.472	ERCC2	ENSG00000104884	NON_SYNONYMOUS_CODING	ENST00000391945.4:c.773G>A	ENSP00000375809.3:p.Arg258Gln	9/23	YES	-	-
S39	SNV	chr19	45867535	C	T	301	159	142	0.472	ERCC2	ENSG00000104884	NON_SYNONYMOUS_CODING	ENST00000586131.1:c.701G>A	ENSP00000464887.1:p.Arg234Gln	9/10	-	-	-
S39	SNV	chr19	45867535	C	T	301	159	142	0.472	ERCC2	ENSG00000104884	NON_SYNONYMOUS_CODING	ENST00000391941.2:c.701G>A	ENSP00000375805.2:p.Arg234Gln	8/21	-	-	-
S32	INDEL	chr2	128029022	T	-C	509	245	264	0.518	ERCC3	ENSG00000163161	FRAME_SHIFT	ENST00000285398.2:c.1835delA	ENSP00000285398.2:p.Asp612AlafsTer29	12/15	YES	-	-
S32	INDEL	chr2	128029022	T	-C	509	245	264	0.518	ERCC3	ENSG00000163161	FRAME_SHIFT	ENST00000493187.2:c.1643delA	ENSP00000447796.1:p.Asp548AlafsTer29	12/15	-	-	-
S20	SNV	chr16	14042064	A	G	1264	682	582	0.46	ERCC4	ENSG00000175595	NON_SYNONYMOUS_CODING	ENST00000311895.7:c.2611A>G	ENSP00000310520.7:p.Lys871Glu	11/11	YES	-	-
31	SNV	chr13	103518074	C	G	304	161	143	0.469	ERCC5	ENSG00000134899	UTR_5_PRIME	ENST00000375954.1:c.-640C>G	-	1/6	-	-	-
31	SNV	chr13	103518074	C	G	304	161	143	0.469	ERCC5	ENSG00000134899	NON_SYNONYMOUS_CODING	ENST00000355739.4:c.2012C>G	ENSP00000347978.4:p.Pro671Arg	9/15	YES	-	-
55	SNV	chr3	185797727	C	G	195	148	47	0.241	ETV5	ENSG00000244405	NON_SYNONYMOUS_CODING	ENST00000537818.1:c.655G>C	ENSP00000441737.1:p.Ala219Pro	7/13	-	0.0008	rs185636057,COSM3846945,COSM1251548,COSM1251547,COSM3846946,COSM3846947
55	SNV	chr3	185797727	C	G	195	148	47	0.241	ETV5	ENSG00000244405	NON_SYNONYMOUS_CODING	ENST00000306376.5:c.529G>C	ENSP00000306376.5:p.Ala177Pro	7/13	YES	0.0008	rs185636057,COSM3846945,COSM1251548,COSM1251547,COSM3846946,COSM3846947
55	SNV	chr3	185797727	C	G	195	148	47	0.241	ETV5	ENSG00000244405	NON_SYNONYMOUS_CODING	ENST00000434744.1:c.529G>C	ENSP00000413755.1:p.Ala177Pro	7/13	-	0.0008	rs185636057,COSM3846945,COSM1251548,COSM1251547,COSM3846946,COSM3846947
S65	INDEL	chr3	185826274	T	+G	349	184	165	0.471	ETV5	ENSG00000244405	FRAME_SHIFT	-	-	1/13	-	-	-
25	SNV	chr12	12037445	G	T	340	190	150	0.441	ETV6	ENSG00000139083	NON_SYNONYMOUS_CODING	ENST00000396373.4:c.1076G>T	ENSP00000379658.3:p.Arg359Leu	6/8	YES	-	-
S33	INDEL	chr12	12006487	A	+T	722	571	151	0.209	ETV6	ENSG00000139083	FRAME_SHIFT	-	-	4/8	YES	-	-
10	SNV	chr7	148529751	C	T	50	38	12	0.24	EZH2	ENSG00000106462	STOP_GAINED	ENST00000460911.1:c.338G>A	ENSP00000419711.1:p.Trp113Ter	4/20	-	-	-
10	SNV	chr7	148529751	C	T	50	38	12	0.24	EZH2	ENSG00000106462	STOP_GAINED	ENST00000476773.1:c.311G>A	ENSP00000419050.1:p.Trp104Ter	4/19	-	-	-
10	SNV	chr7	148506462	G	A	89	59	30	0.337	EZH2	ENSG00000106462	NON_SYNONYMOUS_CODING	ENST00000483967.1:c.2008C>T	ENSP00000419856.1:p.Arg670Cys	18/20	-	-	COSM53005
10	SNV	chr7	148529751	C	T	50	38	12	0.24	EZH2	ENSG00000106462	STOP_GAINED	ENST00000536783.1:c.11G>A	ENSP00000439305.1:p.Trp4Ter	5/12	-	-	-
10	SNV	chr7	148506462	G	A	89	59	30	0.337	EZH2	ENSG00000106462	NON_SYNONYMOUS_CODING	ENST00000359995.2:c.1918C>T	ENSP00000223193.2:p.Arg640Cys	17/19	-	-	COSM53005
10	SNV	chr7	148506462	G	A	89	59	30	0.337	EZH2	ENSG00000106462	NON_SYNONYMOUS_CODING	ENST00000460911.1:c.2035C>T	ENSP00000419711.1:p.Arg679Cys	18/20	-	-	COSM53005

10	SNV	chr7	148506462	G	A	89	59	30	0.337	EZH2	ENSG00000106462	NON_SYNONYMOUS_CODING	ENST00000478654.1:c.1882C>T	ENSP00000417062.1:p.Arg628Cys	18/20	-	-	COSM53005
10	SNV	chr7	148506462	G	A	89	59	30	0.337	EZH2	ENSG00000106462	NON_SYNONYMOUS_CODING	ENST00000541220.1:c.1882C>T	ENSP00000443219.1:p.Arg628Cys	17/19	-	-	COSM53005
10	SNV	chr7	148506462	G	A	89	59	30	0.337	EZH2	ENSG00000106462	NON_SYNONYMOUS_CODING	ENST00000476773.1:c.1882C>T	ENSP00000419050.1:p.Arg628Cys	17/19	-	-	COSM53005
10	SNV	chr7	148506462	G	A	89	59	30	0.337	EZH2	ENSG00000106462	NON_SYNONYMOUS_CODING	ENST00000320356.2:c.2050C>T	ENSP00000320147.2:p.Arg684Cys	18/20	YES	-	COSM53005
10	SNV	chr7	148529751	C	T	50	38	12	0.24	EZH2	ENSG00000106462	STOP_GAINED	ENST00000541220.1:c.311G>A	ENSP00000443219.1:p.Trp104Ter	4/19	-	-	-
10	SNV	chr7	148529751	C	T	50	38	12	0.24	EZH2	ENSG00000106462	STOP_GAINED	ENST00000320356.2:c.338G>A	ENSP00000320147.2:p.Trp113Ter	4/20	YES	-	-
10	SNV	chr7	148529751	C	T	50	38	12	0.24	EZH2	ENSG00000106462	STOP_GAINED	ENST00000483967.1:c.311G>A	ENSP00000419856.1:p.Trp104Ter	4/20	-	-	-
10	SNV	chr7	148529751	C	T	50	38	12	0.24	EZH2	ENSG00000106462	STOP_GAINED	ENST00000478654.1:c.311G>A	ENSP00000417062.1:p.Trp104Ter	5/20	-	-	-
S03	SNV	chr7	148507476	C	T	538	271	267	0.496	EZH2	ENSG00000106462	NON_SYNONYMOUS_CODING	ENST00000350995.2:c.1846G>A	ENSP00000223193.2:p.Gly616Arg	16/19	-	-	COSM53027
S03	SNV	chr7	148504761	C	T	373	191	182	0.488	EZH2	ENSG00000106462	NON_SYNONYMOUS_CODING	ENST00000460911.1:c.2218G>A	ENSP00000419711.1:p.Glu740Lys	20/20	-	-	rs397515548,COSM1087033,COSM1087034
S03	SNV	chr7	148504761	C	T	373	191	182	0.488	EZH2	ENSG00000106462	NON_SYNONYMOUS_CODING	ENST00000476773.1:c.2065G>A	ENSP00000419050.1:p.Glu689Lys	19/19	-	-	rs397515548,COSM1087033,COSM1087034
S03	SNV	chr7	148504761	C	T	373	191	182	0.488	EZH2	ENSG00000106462	NON_SYNONYMOUS_CODING	ENST00000350995.2:c.2101G>A	ENSP00000223193.2:p.Glu701Lys	19/19	-	-	rs397515548,COSM1087033,COSM1087034
S03	SNV	chr7	148507476	C	T	538	271	267	0.496	EZH2	ENSG00000106462	NON_SYNONYMOUS_CODING	ENST00000483967.1:c.1936G>A	ENSP00000419856.1:p.Gly646Arg	17/20	-	-	COSM53027
S03	SNV	chr7	148504761	C	T	373	191	182	0.488	EZH2	ENSG00000106462	NON_SYNONYMOUS_CODING	ENST00000320356.2:c.2233G>A	ENSP00000320147.2:p.Glu745Lys	20/20	YES	-	rs397515548,COSM1087033,COSM1087034
S03	SNV	chr7	148504761	C	T	373	191	182	0.488	EZH2	ENSG00000106462	NON_SYNONYMOUS_CODING	ENST00000483967.1:c.2191G>A	ENSP00000419856.1:p.Glu731Lys	20/20	-	-	rs397515548,COSM1087033,COSM1087034
S03	SNV	chr7	148507476	C	T	538	271	267	0.496	EZH2	ENSG00000106462	NON_SYNONYMOUS_CODING	ENST00000476773.1:c.1810G>A	ENSP00000419050.1:p.Gly604Arg	16/19	-	-	COSM53027
S03	SNV	chr7	148507476	C	T	538	271	267	0.496	EZH2	ENSG00000106462	NON_SYNONYMOUS_CODING	ENST00000541220.1:c.1810G>A	ENSP00000443219.1:p.Gly604Arg	16/19	-	-	COSM53027
S03	SNV	chr7	148504761	C	T	373	191	182	0.488	EZH2	ENSG00000106462	NON_SYNONYMOUS_CODING	ENST00000541220.1:c.2065G>A	ENSP00000443219.1:p.Gly689Lys	19/19	-	-	rs397515548,COSM1087033,COSM1087034
S03	SNV	chr7	148507476	C	T	538	271	267	0.496	EZH2	ENSG00000106462	NON_SYNONYMOUS_CODING	ENST00000460911.1:c.1963G>A	ENSP00000419711.1:p.Gly655Arg	17/20	-	-	COSM53027
S03	SNV	chr7	148504761	C	T	373	191	182	0.488	EZH2	ENSG00000106462	NON_SYNONYMOUS_CODING	ENST00000478654.1:c.2065G>A	ENSP00000417062.1:p.Glu689Lys	20/20	-	-	rs397515548,COSM1087033,COSM1087034
S03	SNV	chr7	148507476	C	T	538	271	267	0.496	EZH2	ENSG00000106462	NON_SYNONYMOUS_CODING	ENST00000320356.2:c.1978G>A	ENSP00000320147.2:p.Gly660Arg	17/20	YES	-	COSM53027
S03	SNV	chr7	148507476	C	T	538	271	267	0.496	EZH2	ENSG00000106462	NON_SYNONYMOUS_CODING	ENST00000478654.1:c.1810G>A	ENSP00000417062.1:p.Gly604Arg	17/20	-	-	COSM53027
S59	SNV	chr7	148514481	C	G	406	222	184	0.453	EZH2	ENSG00000106462	NON_SYNONYMOUS_CODING	ENST00000460911.1:c.1228G>C	ENSP00000419711.1:p.Ala410Pro	11/20	-	-	-
S59	SNV	chr7	148516717	G	A	566	309	257	0.454	EZH2	ENSG00000106462	NON_SYNONYMOUS_CODING	ENST00000460911.1:c.955C>T	ENSP00000419711.1:p.Pro319Ser	9/20	-	-	-
S59	SNV	chr7	148514481	C	G	406	222	184	0.453	EZH2	ENSG00000106462	SPLICE_SITE_REGION	ENST00000460911.1:c.1228G>C	ENSP00000419711.1:p.Ala410Pro	11/20	-	-	-
S59	SNV	chr7	148514481	C	G	406	222	184	0.453	EZH2	ENSG00000106462	SPLICE_SITE_REGION	ENST00000350995.2:c.1111G>C	ENSP00000223193.2:p.Ala371Pro	10/19	-	-	-
S59	SNV	chr7	148514481	C	G	406	222	184	0.453	EZH2	ENSG00000106462	NON_SYNONYMOUS_CODING	ENST00000541220.1:c.1201G>C	ENSP00000443219.1:p.Ala401Pro	11/19	-	-	-
S59	SNV	chr7	148514481	C	G	406	222	184	0.453	EZH2	ENSG00000106462	SPLICE_SITE_REGION	ENST00000320356.2:c.1243G>C	ENSP00000320147.2:p.Ala415Pro	11/20	YES	-	-
S59	SNV	chr7	148514481	C	G	406	222	184	0.453	EZH2	ENSG00000106462	NON_SYNONYMOUS_CODING	ENST00000350995.2:c.1111G>C	ENSP00000223193.2:p.Ala371Pro	10/19	-	-	-
S59	SNV	chr7	148516717	G	A	566	309	257	0.454	EZH2	ENSG00000106462	NON_SYNONYMOUS_CODING	ENST00000483967.1:c.928C>T	ENSP00000419856.1:p.Pro310Ser	9/20	-	-	-
S59	SNV	chr7	148516717	G	A	566	309	257	0.454	EZH2	ENSG00000106462	NON_SYNONYMOUS_CODING	ENST00000541220.1:c.928C>T	ENSP00000443219.1:p.Pro310Ser	9/19	-	-	-
S59	SNV	chr7	148514481	C	G	406	222	184	0.453	EZH2	ENSG00000106462	SPLICE_SITE_REGION	ENST00000541220.1:c.1201G>C	ENSP00000443219.1:p.Ala401Pro	11/19	-	-	-
S59	SNV	chr7	148516717	G	A	566	309	257	0.454	EZH2	ENSG00000106462	NON_SYNONYMOUS_CODING	ENST00000478654.1:c.928C>T	ENSP00000417062.1:p.Pro310Ser	10/20	-	-	-
S59	SNV	chr7	148514481	C	G	406	222	184	0.453	EZH2	ENSG00000106462	SPLICE_SITE_REGION	ENST00000478654.1:c.1201G>C	ENSP00000417062.1:p.Ala401Pro	12/20	-	-	-
S59	SNV	chr7	148516717	G	A	566	309	257	0.454	EZH2	ENSG00000106462	NON_SYNONYMOUS_CODING	ENST00000350995.2:c.838C>T	ENSP00000223193.2:p.Pro280Ser	8/19	-	-	-
S59	SNV	chr7	148514481	C	G	406	222	184	0.453	EZH2	ENSG00000106462	SPLICE_SITE_REGION	ENST00000476773.1:c.1201G>C	ENSP00000419050.1:p.Ala401Pro	11/19	-	-	-
S59	SNV	chr7	148516717	G	A	566	309	257	0.454	EZH2	ENSG00000106462	NON_SYNONYMOUS_CODING	ENST00000320356.2:c.970C>T	ENSP00000320147.2:p.Pro324Ser	9/20	YES	-	-
S59	SNV	chr7	148514481	C	G	406	222	184	0.453	EZH2	ENSG00000106462	NON_SYNONYMOUS_CODING	ENST00000320356.2:c.1243G>C	ENSP00000320147.2:p.Ala415Pro	11/20	YES	-	-
S59	SNV	chr7	148514481	C	G	406	222	184	0.453	EZH2	ENSG00000106462	SPLICE_SITE_REGION	ENST00000483967.1:c.1201G>C	ENSP00000419856.1:p.Ala401Pro	11/20	-	-	-
S59	SNV	chr7	148514481	C	G	406	222	184	0.453	EZH2	ENSG00000106462	NON_SYNONYMOUS_CODING	ENST00000476773.1:c.1201G>C	ENSP00000419050.1:p.Ala401Pro	11/19	-	-	-
S59	SNV	chr7	148514481	C	G	406	222	184	0.453	EZH2	ENSG00000106462	NON_SYNONYMOUS_CODING	ENST00000478654.1:c.1201G>C	ENSP00000417062.1:p.Ala401Pro	12/20	-	-	-
S59	SNV	chr7	148516717	G	A	566	309	257	0.454	EZH2	ENSG00000106462	NON_SYNONYMOUS_CODING	ENST00000536783.1:c.628C>T	ENSP00000439305.1:p.Pro210Ser	10/12	-	-	-
S59	SNV	chr7	148516717	G	A	566	309	257	0.454	EZH2	ENSG00000106462	NON_SYNONYMOUS_CODING	ENST00000476773.1:c.928C>T	ENSP00000419050.1:p.Pro310Ser	9/19	-	-	-
S59	SNV	chr7	148514481	C	G	406	222	184	0.453	EZH2	ENSG00000106462	NON_SYNONYMOUS_CODING	ENST00000483967.1:c.1201G>C	ENSP00000419856.1:p.Ala401Pro	11/20	-	-	-
S38	SNV	chr6	159239116	G	A	534	249	285	0.534	EZR	ENSG00000092820	NON_SYNONYMOUS_CODING	ENST00000337147.7:c.10C>T	ENSP00000338934.7:p.Pro4Ser	1/13	-	-	-
S38	SNV	chr6	159239116	G	A	534	249	285	0.534	EZR	ENSG00000092820	SPLICE_SITE_REGION	ENST00000392177.4:c.10C>T	ENSP00000376016.4:p.Pro4Ser	1/12	-	-	-
S38	SNV	chr6	159239116	G	A	534	249	285	0.534	EZR	ENSG00000092820	SPLICE_SITE_REGION	ENST00000337147.7:c.10C>T	ENSP00000338934.7:p.Pro4Ser	1/13	-	-	-
S38	SNV	chr6	159239116	G	A	534	249	285	0.534	EZR	ENSG00000092820	NON_SYNONYMOUS_CODING	ENST00000392177.4:c.10C>T	ENSP00000376016.4:p.Pro4Ser	1/12	-	-	-
S38	SNV	chr6	159239116	G	A	534	249	285	0.534	EZR	ENSG00000092820	NON_SYNONYMOUS_CODING	ENST00000367075.3:c.10C>T	ENSP00000356042.3:p.Pro4Ser	2/14	YES	-	-
S38	SNV	chr6	159239116	G	A	534	249	285	0.534	EZR	ENSG00000092820	SPLICE_SITE_REGION	ENST00000367075.3:c.10C>T	ENSP00000356042.3:p.Pro4Ser	2/14	YES	-	-
11	SNV	chr16	89837018	A	C	59	43	16	0.271	FANCA	ENSG00000187741	NON_SYNONYMOUS_CODING	ENST00000389301.3:c.2176T>G	ENSP00000373952.3:p.Phe726Val	24/43	YES	-	-
S47	SNV	chr16	89874736	G	C	386	218	168	0.435	FANCA	ENSG00000187741	NON_SYNONYMOUS_CODING	ENST00000389301.3:c.562C>G	ENSP00000373952.3:p.Gln188Glu	6/43	YES	-	-
S47	SNV	chr16	89874736	G	C	386	218	168	0.435	FANCA	ENSG00000187741	NON_SYNONYMOUS_CODING	ENST00000534992.1:c.562C>G	ENSP00000443675.1:p.Gln188Glu	6/11	-	-	-
S47	SNV	chr16	89874736	G	C	386	218	168	0.435	FANCA	ENSG00000187741	NON_SYNONYMOUS_CODING	ENST00000567673.1:c.562C>G	ENSP00000456443.1:p.Gln188Glu	6/10	-	-	-
S47	SNV	chr16	89874736	G	C	386	218	168	0.435	FANCA	ENSG00000187741	NON_SYNONYMOUS_CODING	ENST00000543736.1:c.466C>G	ENSP00000443409.1:p.Gln156Glu	5/10	-	-	-
S47	SNV	chr16	89874736	G	C	386	218	168	0.435	FANCA	ENSG00000187741	NON_SYNONYMOUS_CODING	ENST00000568369.1:c.562C>G	ENSP00000456829.1:p.Gln188Glu	6/43	-	-	-
S47	SNV	chr16	89874736	G	C	386	218	168	0.435	FANCA	ENSG00000187741	NON_SYNONYMOUS_CODING	ENST00000389302.3:c.562C>G	ENSP00000373953.3:p.Gln188Glu	6/11	-	-	-
1	SNV	chr9	97933410	C	T	194	111	83	0.428	FANCC	ENSG00000158169	NON_SYNONYMOUS_CODING	ENST00000289081.3:c.472G>A	ENSP00000289081.3:p.Ala158Thr	6/15	YES	-	rs372338418
1	SNV	chr9	98011619	A	G	183	98	85	0.464	FANCC	ENSG00000158169	UTR_5_PRIME	ENST00000375305.1:c.-46T>C	-	2/15	-	-	-
1	SNV	chr9	97933410	C	T	194	111	83	0.428	FANCC	ENSG00000158169	NON_SYNONYMOUS_CODING	ENST00000375305.1:c.472G>A	ENSP00000364454.1:p.Ala158Thr	6/15	-	-	rs372338418
1	SNV	chr9	98011619	A	G	183	98	85	0.464	FANCC	ENSG00000158169	UTR_5_PRIME	ENST00000289081.3:c.-46T>C	-	2/15	YES	-	-
1	SNV	chr9	98011619	A	G	183	98	85	0.464	FANCC	ENSG00000158169	UTR_5_PRIME	ENST00000433829.1:c.-46T>C	-	2/5	-	-	-
21	SNV	chr9	97897632	G	A	230	123	107	0.465	FANCC	ENSG00000158169	NON_SYNONYMOUS_CODING	ENST00000375305.1:c.839C>T	ENSP00000364454.1:p.Ser280Leu	8/15	-	-	-
21	SNV	chr9	97897632	G	A	230	123	107	0.465	FANCC	ENSG00000158169	NON_SYNONYMOUS_CODING	ENST00000289081.3:c.839C>T	ENSP00000289081.3:p.Ser280Leu	8/15	YES	-	-
S60	SNV	chr9	97897732	G	A	641	338	303	0.473	FANCC	ENSG00000158169	NON_SYNONYMOUS_CODING	ENST00000375305.1:c.739C>T	ENSP00000364454.1:p.Leu247Phe	8/15	-	-	-
S60	SNV	chr9	97897732	G	A	641	338	303	0.473	FANCC	ENSG00000158169	NON_SYNONYMOUS_CODING	ENST00000289081.3:c.739C>T	ENSP00000289081.3:p.Leu247Phe	8/15	YES	-	-
31	SNV	chr11	22647134	C	T	256	144	112	0.438	FANCF	ENSG00000183161	NON_SYNONYMOUS_CODING	ENST00000327470.3:c.223G>A	ENSP00000330875.3:p.Gly75Ser	1/1	YES	-	-
24	SNV	chr4	187518126	C	T	411	265	146	0.355	FAT1	ENSG00000083857	NON_SYNONYMOUS_CODING	ENST00000441802.2:c.12568G>A	ENSP00000406229.2:p.Ala4190Thr	25/27	YES	-	-
31	SNV	chr4	187627794	T	C	200	127	133	0.511	FAT1	ENSG00000083857	NON_SYNONYMOUS_CODING	ENST00000441802.2:c.3248A>G	ENSP00000406229.2:p.Lys1083Arg	2/27	YES	-	-
64	SNV	chr4	187538299	A	G	95	50	45	0.474	FAT1	ENSG00000083857	NON_SYNONYMOUS_CODING	ENST00000441802.2:c.8935T>C	ENSP00000406229.2:p.Tyr2979His	11/27	YES	-	-
S19	SNV	chr4	187509919	C	T	537	278	259	0.482	FAT1	ENSG00000083857	NON_SYNONYMOUS_CODING	ENST00000512772.1:c.932G>A	ENSP00000424157.1:p.Val312Ile	4/4	-	-	-
S19	SNV	chr4	187509919	C	T	537	278	259	0.482	FAT1	ENSG00000083857	NON_SYNONYMOUS_CODING	ENST00000441802.2:c.13594G>A	ENSP00000406229.2:p.Val4532Ile	27/27	YES	-	-
S38	SNV	chr4	187541862	C	G	701	362	339	0.484	FAT1	ENSG00000083857	NON_SYNONYMOUS_CODING	ENST00000441802.2:c.5878G>C	ENSP00000406229.2:p.Ala1960Pro	10/27	YES	-	-
S46	SNV	chr4	187628169	T	C	613	321	292	0.476	FAT1	ENSG00000083857	NON_SYNONYMOUS_CODING	ENST00000441802.2:c.2813A>G	ENSP00000406229.2:p.Glu938Gly	2/27	YES	-	-
21	SNV	chr5	150947190	T	C	606	347	259	0.427	FAT2	ENSG00000086570	NON_SYNONYMOUS_CODING	ENST00000261800.5:c.1303A>G	ENSP00000261800.5:p.Thr435Ala	1/23	YES	-	-
22	SNV	chr5	150933988	C	T	659	330	329	0.499	FAT2	ENSG00000086570	NON_SYNONYMOUS_CODING	ENST00000261800.5:c.3880G>A	ENSP00000261800.5:p.Asp1294Asn	4/23	YES	-	-
S65	SNV	chr5	150924104	G	A	821	422	399	0.486	FAT2	ENSG00000086570	NON_SYNONYMOUS_CODING	ENST00000261800.5:c.6584C>T	ENSP00000261800.5:p.Ala2195Val	9/23	YES	-	-
12	SNV	chr11	92531596	A	G	160	95	65	0.406	FAT3	ENSG00000165323	NON_SYNONYMOUS_CODING	ENST00000298047.6:c.5417A>G	ENSP00000298047.6:p.Asn1806Ser	9/27	YES	-	-
12	SNV	chr11	92531596	A	G	160	95	65	0.406	FAT3	ENSG00000165323	NON_SYNONYMOUS_CODING	ENST00000409404.2:c.5417A>G	ENSP00000387040.2:p.Asn1806Ser	9/25	-	-	-
12	SNV	chr11	92531596	A	G	160	95	65	0.406	FAT3	ENSG00000165323	NON_SYNONYMOUS_CODING	ENST00000525166.1:c.4967A>G	ENSP00000432586.1:p.Asn1656Ser	9/27	-	-	-
S61	SNV	chr11	92565100	T	C	845	440	405	0.479	FAT3	ENSG00000165323	NON_SYNONYMOUS_CODING	ENST00000525166.1:c.9344T>C	ENSP00000432586.1:p.Ile3115Thr	13/27	-	-	-
S61	SNV	chr11	92565100	T	C	845	440	405	0.479	FAT3	ENSG00000165323	NON_SYNONYMOUS_CODING	ENST00000409404.2:c.9794T>C	ENSP00000387040.2:p.Ile3265Thr	13/25	-	-	-
S61	SNV	chr11	92565100	T	C	845	440	405	0.479	FAT3	ENSG00000165323	NON_SYNONYMOUS_CODING	ENST00000298047.6:c.9794T>C	ENSP00000298047.6:p.Ile3265Thr	13/27	YES	-	-
59	INDEL	chr4	153332910	C	+AGG	32	20	12	0.375	FBXW7	ENSG00000109670	CODON_INSERTION	-	-	2/12	YES	-	rs368702202
S35	SNV	chr1	157557280	G	T	525	260	265	0.505	FCRL4	ENSG00000163518	NON_SYNONYMOUS_CODING	ENST00000271532.1:c.833C>A	ENSP00000271532.1:p.Ser211Arg	5/12	YES	-	-
10	SNV	chr10	123263407	T	C	187	99	88	0.468	FGFR2	ENSG00000066468	NON_SYNONYMOUS_CODING	ENST00000369060.4:c.988A>G	ENSP00000358056.4:p.Arg330Gly	8/16	-	-	-
10	SNV	chr10	123263407	T	C	187	99	88	0.468	FGFR2	ENSG00000066468	NON_SYNONYMOUS_CODING	ENST00000360144.3:c.1072A>G	ENSP00000353262.3:p.Arg358Gly	9/17	-	-	-
10	SNV	chr10	123263407	T	C	187	99	88	0.468	FGFR2	ENSG00000066468	NON_SYNONYMOUS_CODING	ENST00000358487.5:c.1336A>G	ENSP00000351276.5:p.Arg446Gly	10/18	-	-	-
10	SNV	chr10	123263407	T	C	187	99	88	0.468	FGFR2	ENSG00000066468	NON_SYNONYMOUS_CODING	ENST00000457416.2:c.1339A>G	ENSP00000410294.2:p.Arg447Gly	10/18	YES	-	-
10	SNV	chr10	123263407	T	C	187	99	88	0.468	FGFR2	ENSG00000066468	NON_SYNONYMOUS_CODING	ENST00000336553.6:c.1063A>G	ENSP00000337665.6:p.Arg355Gly	9/16	-	-	-
10	SNV	chr10	123263407	T	C	187	99	88	0.468	FGFR2	ENSG00000066468	NON_SYNONYMOUS_CODING	ENST00000478659.1:c.652A>G	ENSP00000474013.1:p.Arg218Gly	9/17	-	-	-
10	SNV	chr10	123263407	T	C	187	99	88	0.468	FGFR2	ENSG00000066468	NON_SYNONYMOUS_CODING	ENST00000369056.1:c.1339A>G	ENSP00000358052.1:p.Arg447Gly	9/17	-	-	-
10	SNV	chr10	123263407	T	C	187	99	88	0.468	FGFR2	ENSG00000066468	NON_SYNONYMOUS_CODING	ENST00000429361.1:c.112A>G	ENSP00000404219.1:p.Arg38Gly	2/9	-	-	-
10	SNV	chr10	123263407	T	C	187	99	88	0.468	FGFR2	ENSG00000066468	NON_SYNONYMOUS_CODING	ENST00000351936.6:c.1330A>G	ENSP00000330987.8:p.Arg444Gly	10/18	-	-	-
10	SNV	chr10	123263407	T	C	187	99	88	0.468	FGFR2	ENSG00000066468	NON_SYNONYMOUS_CODING	ENST00000357555.5:c.1069A>G	ENSP00000350166.5:p.Arg357Gly	9/17	-	-	-
10	SNV	chr10	123263407	T	C	187	99	88	0.468	FGFR2	ENSG00000066468	NON_SYNONYMOUS_CODING	ENST00000369059.1:c.994A>G	ENSP00000358055.1:p.Arg332Gly	8/16	-	-	-
10	SNV	chr10	123263407	T	C	187	99	88	0.468	FGFR2	ENSG00000066468	NON_SYNONYMOUS_CODING	ENST00000369058.3:c.1339A>G	ENSP00000358054.3:p.Arg447Gly	10/17	-	-	-
10	SNV	chr10	123263407	T	C	187	99	88	0.468	FGFR2	ENSG00000066468	NON_SYNONYMOUS_CODING	ENST00000356226.4:c.985A>G	ENSP00000348559.4:p.Arg329Gly	8/16	-	-	-
10	SNV	chr10	123263407	T	C	187	99	88	0.468	FGFR2	ENSG00000066468	NON_SYNONYMOUS_CODING	ENST00000369061.4:c.1000A>G	ENSP00000358057.4:p.Arg334Gly	7/15	-	-	-

10	SNV	chr10	123263407	T	C	187	99	88	0.468	FGFR2	ENSG00000066468	NON_SYNONYMOUS_CODING	ENST00000346997.2:c.1330A>G	ENSP00000263451.5:p.Arg444Gly	9/17	-	-	-
38	SNV	chr10	123260370	C	T	519	251	268	0.515	FGFR2	ENSG00000066468	NON_SYNONYMOUS_CODING	ENST00000429361.1:c.307G>A	ENSP00000404219.1:p.Ala103Thr	3/9	-	-	-
38	SNV	chr10	123260370	C	T	519	251	268	0.515	FGFR2	ENSG00000066468	NON_SYNONYMOUS_CODING	ENST00000369059.1:c.1189G>A	ENSP00000358055.1:p.Ala397Thr	9/16	-	-	-
38	SNV	chr10	123260370	C	T	519	251	268	0.515	FGFR2	ENSG00000066468	NON_SYNONYMOUS_CODING	ENST00000358487.5:c.1531G>A	ENSP00000351276.5:p.Ala511Thr	11/18	-	-	-
38	SNV	chr10	123260370	C	T	519	251	268	0.515	FGFR2	ENSG00000066468	NON_SYNONYMOUS_CODING	ENST00000369061.4:c.1195G>A	ENSP00000358057.4:p.Ala399Thr	8/15	-	-	-
38	SNV	chr10	123260370	C	T	519	251	268	0.515	FGFR2	ENSG00000066468	NON_SYNONYMOUS_CODING	ENST00000357555.5:c.1264G>A	ENSP00000350166.5:p.Ala422Thr	10/17	-	-	-
38	SNV	chr10	123260370	C	T	519	251	268	0.515	FGFR2	ENSG00000066468	NON_SYNONYMOUS_CODING	ENST00000336553.6:c.1258G>A	ENSP00000337665.6:p.Ala420Thr	10/16	-	-	-
38	SNV	chr10	123260370	C	T	519	251	268	0.515	FGFR2	ENSG00000066468	NON_SYNONYMOUS_CODING	ENST00000478859.1:c.847G>A	ENSP00000474011.1:p.Ala283Thr	10/17	-	-	-
38	SNV	chr10	123260370	C	T	519	251	268	0.515	FGFR2	ENSG00000066468	NON_SYNONYMOUS_CODING	ENST00000369060.4:c.1183G>A	ENSP00000358056.4:p.Ala395Thr	9/16	-	-	-
38	SNV	chr10	123260370	C	T	519	251	268	0.515	FGFR2	ENSG00000066468	NON_SYNONYMOUS_CODING	ENST00000457416.2:c.1534G>A	ENSP00000410294.2:p.Ala512Thr	11/18	YES	-	-
38	SNV	chr10	123260370	C	T	519	251	268	0.515	FGFR2	ENSG00000066468	NON_SYNONYMOUS_CODING	ENST00000346997.2:c.1525G>A	ENSP00000263451.5:p.Ala509Thr	10/17	-	-	-
38	SNV	chr10	123260370	C	T	519	251	268	0.515	FGFR2	ENSG00000066468	NON_SYNONYMOUS_CODING	ENST00000360144.3:c.1267G>A	ENSP00000353262.3:p.Ala423Thr	10/17	-	-	-
38	SNV	chr10	123260370	C	T	519	251	268	0.515	FGFR2	ENSG00000066468	NON_SYNONYMOUS_CODING	ENST00000369056.1:c.1534G>A	ENSP00000358052.1:p.Ala512Thr	10/17	-	-	-
38	SNV	chr10	123260370	C	T	519	251	268	0.515	FGFR2	ENSG00000066468	NON_SYNONYMOUS_CODING	ENST00000351936.6:c.1525G>A	ENSP00000309878.8:p.Ala509Thr	11/18	-	-	-
38	SNV	chr10	123260370	C	T	519	251	268	0.515	FGFR2	ENSG00000066468	NON_SYNONYMOUS_CODING	ENST00000356226.4:c.1180G>A	ENSP00000348559.4:p.Ala394Thr	9/16	-	-	-
38	SNV	chr10	123260370	C	T	519	251	268	0.515	FGFR2	ENSG00000066468	NON_SYNONYMOUS_CODING	ENST00000369058.3:c.1534G>A	ENSP00000358054.3:p.Ala512Thr	11/17	-	-	-
58	SNV	chr10	123325039	C	T	375	182	193	0.513	FGFR2	ENSG00000066468	NON_SYNONYMOUS_CODING	ENST00000369061.4:c.289G>A	ENSP00000358057.4:p.Ala97Thr	2/15	-	-	rs372430349,COSM49174
58	SNV	chr10	123325039	C	T	375	182	193	0.513	FGFR2	ENSG00000066468	NON_SYNONYMOUS_CODING	ENST00000359354.2:c.289G>A	ENSP00000352309.2:p.Ala97Thr	3/7	-	-	rs372430349,COSM49174
58	SNV	chr10	123325039	C	T	375	182	193	0.513	FGFR2	ENSG00000066468	NON_SYNONYMOUS_CODING	ENST00000351936.6:c.289G>A	ENSP00000309878.8:p.Ala97Thr	3/18	-	-	rs372430349,COSM49174
58	SNV	chr10	123325039	C	T	375	182	193	0.513	FGFR2	ENSG00000066468	NON_SYNONYMOUS_CODING	ENST00000346997.2:c.289G>A	ENSP00000263451.5:p.Ala97Thr	2/17	-	-	rs372430349,COSM49174
58	SNV	chr10	123325039	C	T	375	182	193	0.513	FGFR2	ENSG00000066468	NON_SYNONYMOUS_CODING	ENST00000369056.1:c.289G>A	ENSP00000358052.1:p.Ala97Thr	2/17	-	-	rs372430349,COSM49174
58	SNV	chr10	123325039	C	T	375	182	193	0.513	FGFR2	ENSG00000066468	NON_SYNONYMOUS_CODING	ENST00000369058.3:c.289G>A	ENSP00000358054.3:p.Ala97Thr	3/17	-	-	rs372430349,COSM49174
58	SNV	chr10	123325039	C	T	375	182	193	0.513	FGFR2	ENSG00000066468	NON_SYNONYMOUS_CODING	ENST00000358487.5:c.289G>A	ENSP00000351276.5:p.Ala97Thr	3/18	-	-	rs372430349,COSM49174
58	SNV	chr10	123325039	C	T	375	182	193	0.513	FGFR2	ENSG00000066468	NON_SYNONYMOUS_CODING	ENST00000457416.2:c.289G>A	ENSP00000410294.2:p.Ala97Thr	3/18	YES	-	rs372430349,COSM49174
58	SNV	chr10	123325039	C	T	375	182	193	0.513	FGFR2	ENSG00000066468	NON_SYNONYMOUS_CODING	ENST00000369060.4:c.289G>A	ENSP00000358056.4:p.Ala97Thr	3/16	-	-	rs372430349,COSM49174
S65	SNV	chr4	1806624	A	T	170	82	88	0.518	FGFR3	ENSG00000068078	NON_SYNONYMOUS_CODING	ENST00000412135.2:c.1004A>T	ENSP00000412903.2:p.Glu335Val	8/16	-	-	-
S65	SNV	chr4	1806623	G	T	168	82	86	0.512	FGFR3	ENSG00000068078	STOP_GAINED	ENST00000340107.4:c.1345G>T	ENSP00000339824.4:p.Glu449Ter	10/18	YES	-	rs35317612
S65	SNV	chr4	1806623	G	T	168	82	86	0.512	FGFR3	ENSG00000068078	STOP_GAINED	ENST00000440486.2:c.1339G>T	ENSP00000414914.2:p.Glu447Ter	10/18	-	-	rs35317612
S65	SNV	chr4	1806623	G	T	168	82	86	0.512	FGFR3	ENSG00000068078	STOP_GAINED	ENST00000481110.2:c.1342G>T	ENSP00000420533.2:p.Glu448Ter	10/17	-	-	rs35317612
S65	SNV	chr4	1806624	A	T	170	82	88	0.518	FGFR3	ENSG00000068078	NON_SYNONYMOUS_CODING	ENST00000340107.4:c.1346A>T	ENSP00000339824.4:p.Glu449Val	10/18	YES	-	-
S65	SNV	chr4	1806623	G	T	168	82	86	0.512	FGFR3	ENSG00000068078	STOP_GAINED	ENST00000260795.2:c.1339G>T	ENSP00000260795.2:p.Glu447Ter	9/17	-	-	rs35317612
S65	SNV	chr4	1806623	G	T	168	82	86	0.512	FGFR3	ENSG00000068078	STOP_GAINED	ENST00000412135.2:c.1003G>T	ENSP00000412903.2:p.Glu335Ter	8/16	-	-	rs35317612
S65	SNV	chr4	1806624	A	T	170	82	88	0.518	FGFR3	ENSG00000068078	NON_SYNONYMOUS_CODING	ENST00000260795.2:c.1340A>T	ENSP00000260795.2:p.Lys447Val	9/17	-	-	-
S65	SNV	chr4	1806624	A	T	170	82	88	0.518	FGFR3	ENSG00000068078	NON_SYNONYMOUS_CODING	ENST00000481110.2:c.1343A>T	ENSP00000420533.2:p.Glu448Val	10/17	-	-	-
S65	SNV	chr4	1806624	A	T	170	82	88	0.518	FGFR3	ENSG00000068078	NON_SYNONYMOUS_CODING	ENST00000352904.1:c.1004A>T	ENSP00000023803.1:p.Glu335Val	7/15	-	-	-
S65	SNV	chr4	1806624	A	T	170	82	88	0.518	FGFR3	ENSG00000068078	NON_SYNONYMOUS_CODING	ENST00000440486.2:c.1340A>T	ENSP00000414914.2:p.Lys447Val	10/18	-	-	-
S65	SNV	chr4	1806623	G	T	168	82	86	0.512	FGFR3	ENSG00000068078	STOP_GAINED	ENST00000352904.1:c.1003G>T	ENSP00000023803.1:p.Glu335Ter	7/15	-	-	rs35317612
1	SNV	chr1	241661075	G	A	92	47	45	0.489	FH	ENSG00000091483	UTR_3_PRIME	ENST00000366560.3:c.*53C>T		10/10	YES	-	-
33	SNV	chrX	153595129	C	T	136	0	136	1	FLNA	ENSG00000196924	NON_SYNONYMOUS_CODING	ENST00000422373.1:c.958G>A	ENSP00000416926.1:p.Val320Met	6/47	-	-	-
33	SNV	chrX	153595129	C	T	136	0	136	1	FLNA	ENSG00000196924	NON_SYNONYMOUS_CODING	ENST00000360319.4:c.958G>A	ENSP00000353467.4:p.Val320Met	5/46	-	-	-
33	SNV	chrX	153595129	C	T	136	0	136	1	FLNA	ENSG00000196924	NON_SYNONYMOUS_CODING	ENST00000344736.4:c.958G>A	ENSP00000358863.3:p.Val320Met	5/45	-	-	-
33	SNV	chrX	153595129	C	T	136	0	136	1	FLNA	ENSG00000196924	NON_SYNONYMOUS_CODING	ENST00000369850.3:c.958G>A	ENSP00000358866.3:p.Val320Met	6/48	YES	-	-
19	SNV	chr13	28592642	C	A	756	407	349	0.462	FLT3	ENSG00000122025	NON_SYNONYMOUS_CODING	ENST00000241453.7:c.2503G>T	ENSP00000241453.7:p.Asp835Tyr	20/24	YES	-	KinMutBase_FLT3_DNA:g.53109G>T,KinMutBase_FLT3_DNA:g.53109G>C,KinMutBase_FLT3_DNA:g.53109G>A,KinMutBase_FLT3_DNA:g.53109delG,COSM783,COSM789,COSM785
19	SNV	chr13	28592642	C	A	756	407	349	0.462	FLT3	ENSG00000122025	NON_SYNONYMOUS_CODING	ENST00000380982.4:c.2503G>T	ENSP00000370369.4:p.Asp835Tyr	20/24	-	-	KinMutBase_FLT3_DNA:g.53109G>T,KinMutBase_FLT3_DNA:g.53109G>C,KinMutBase_FLT3_DNA:g.53109G>A,KinMutBase_FLT3_DNA:g.53109delG,COSM783,COSM789,COSM785
6	SNV	chr13	28592642	C	G	872	525	347	0.386	FLT3	ENSG00000122025	NON_SYNONYMOUS_CODING	ENST00000241453.7:c.2503G>C	ENSP00000241453.7:p.Asp835His	20/24	YES	-	KinMutBase_FLT3_DNA:g.53109G>T,KinMutBase_FLT3_DNA:g.53109G>C,KinMutBase_FLT3_DNA:g.53109G>A,KinMutBase_FLT3_DNA:g.53109delG,COSM783,COSM789,COSM785
6	SNV	chr13	28592642	C	G	872	525	347	0.386	FLT3	ENSG00000122025	NON_SYNONYMOUS_CODING	ENST00000380982.4:c.2503G>C	ENSP00000370369.4:p.Asp835His	20/24	-	-	KinMutBase_FLT3_DNA:g.53109G>T,KinMutBase_FLT3_DNA:g.53109G>C,KinMutBase_FLT3_DNA:g.53109G>A,KinMutBase_FLT3_DNA:g.53109delG,COSM783,COSM789,COSM785
7	SNV	chr13	28608276	A	T	349	244	105	0.301	FLT3	ENSG00000122025	NON_SYNONYMOUS_CODING	ENST00000380982.4:c.1780T>A	ENSP00000370369.4:p.Phe594Ile	14/24	-	-	COSM96544
7	SNV	chr13	28608276	A	T	349	244	105	0.301	FLT3	ENSG00000122025	NON_SYNONYMOUS_CODING	ENST00000241453.7:c.1780T>A	ENSP00000241453.7:p.Phe594Ile	14/24	YES	-	COSM96544
7	SNV	chr13	28608276	A	T	349	244	105	0.301	FLT3	ENSG00000122025	NON_SYNONYMOUS_CODING	ENST00000537084.1:c.1780T>A	ENSP00000438139.1:p.Phe594Ile	14/23	-	-	COSM96544
S03	SNV	chr13	28592642	C	A	665	409	256	0.385	FLT3	ENSG00000122025	NON_SYNONYMOUS_CODING	ENST00000380982.4:c.2503G>T	ENSP00000370369.4:p.Asp835Tyr	20/24	-	-	KinMutBase_FLT3_DNA:g.53109G>T,KinMutBase_FLT3_DNA:g.53109G>C,KinMutBase_FLT3_DNA:g.53109G>A,KinMutBase_FLT3_DNA:g.53109delG,COSM783,COSM789,COSM785
S03	SNV	chr13	28592642	C	A	665	409	256	0.385	FLT3	ENSG00000122025	NON_SYNONYMOUS_CODING	ENST00000241453.7:c.2503G>T	ENSP00000241453.7:p.Asp835Tyr	20/24	YES	-	KinMutBase_FLT3_DNA:g.53109G>T,KinMutBase_FLT3_DNA:g.53109G>C,KinMutBase_FLT3_DNA:g.53109G>A,KinMutBase_FLT3_DNA:g.53109delG,COSM783,COSM789,COSM785
S12	SNV	chr13	28592642	C	G	684	388	296	0.433	FLT3	ENSG00000122025	NON_SYNONYMOUS_CODING	ENST00000241453.7:c.2503G>C	ENSP00000241453.7:p.Asp835His	20/24	YES	-	KinMutBase_FLT3_DNA:g.53109G>T,KinMutBase_FLT3_DNA:g.53109G>C,KinMutBase_FLT3_DNA:g.53109G>A,KinMutBase_FLT3_DNA:g.53109delG,COSM783,COSM789,COSM785
S12	SNV	chr13	28592642	C	G	684	388	296	0.433	FLT3	ENSG00000122025	NON_SYNONYMOUS_CODING	ENST00000380982.4:c.2503G>C	ENSP00000370369.4:p.Asp835His	20/24	-	-	KinMutBase_FLT3_DNA:g.53109G>T,KinMutBase_FLT3_DNA:g.53109G>C,KinMutBase_FLT3_DNA:g.53109G>A,KinMutBase_FLT3_DNA:g.53109delG,COSM783,COSM789,COSM785
S32	SNV	chr13	28592642	C	G	682	512	170	0.214	FLT3	ENSG00000122025	NON_SYNONYMOUS_CODING	ENST00000241453.7:c.2503G>C	ENSP00000241453.7:p.Asp835His	20/24	YES	-	KinMutBase_FLT3_DNA:g.53109G>T,KinMutBase_FLT3_DNA:g.53109G>C,KinMutBase_FLT3_DNA:g.53109G>A,KinMutBase_FLT3_DNA:g.53109delG,COSM783,COSM789,COSM785
S32	SNV	chr13	28592642	C	G	682	512	170	0.214	FLT3	ENSG00000122025	NON_SYNONYMOUS_CODING	ENST00000380982.4:c.2503G>C	ENSP00000370369.4:p.Asp835His	20/24	-	-	KinMutBase_FLT3_DNA:g.53109G>T,KinMutBase_FLT3_DNA:g.53109G>C,KinMutBase_FLT3_DNA:g.53109G>A,KinMutBase_FLT3_DNA:g.53109delG,COSM783,COSM789,COSM785
S48	SNV	chr13	28608281	A	T	481	304	177	0.368	FLT3	ENSG00000122025	NON_SYNONYMOUS_CODING	ENST00000537084.1:c.1775T>A	ENSP00000438139.1:p.Val592Asp	14/23	-	-	COSM28045,COSM19522
S48	SNV	chr13	28608281	A	T	481	304	177	0.368	FLT3	ENSG00000122025	NON_SYNONYMOUS_CODING	ENST00000380982.4:c.1775T>A	ENSP00000370369.4:p.Val592Asp	14/24	-	-	COSM28045,COSM19522
S48	SNV	chr13	28608281	A	T	481	304	177	0.368	FLT3	ENSG00000122025	NON_SYNONYMOUS_CODING	ENST00000241453.7:c.1775T>A	ENSP00000241453.7:p.Val592Asp	14/24	YES	-	COSM28045,COSM19522
S65	SNV	chr13	28592642	C	A	794	589	205	0.258	FLT3	ENSG00000122025	NON_SYNONYMOUS_CODING	ENST00000241453.7:c.2503G>T	ENSP00000241453.7:p.Asp835Tyr	20/24	YES	-	KinMutBase_FLT3_DNA:g.53109G>T,KinMutBase_FLT3_DNA:g.53109G>C,KinMutBase_FLT3_DNA:g.53109G>A,KinMutBase_FLT3_DNA:g.53109delG,COSM783,COSM789,COSM785
S65	SNV	chr13	28592642	C	A	794	589	205	0.258	FLT3	ENSG00000122025	NON_SYNONYMOUS_CODING	ENST00000380982.4:c.2503G>T	ENSP00000370369.4:p.Asp835Tyr	20/24	-	-	KinMutBase_FLT3_DNA:g.53109G>T,KinMutBase_FLT3_DNA:g.53109G>C,KinMutBase_FLT3_DNA:g.53109G>A,KinMutBase_FLT3_DNA:g.53109delG,COSM783,COSM789,COSM785
S47	INDEL	chr3	138664890	C	-GCT	213	101	112	0.526	FOXL2	ENSG00000183770	CODON_DELETION	ENST00000330315.3:c.676delG	ENSP00000333188.3:p.Ala226LeufsTer45	1/1	YES	-	-
S66	INDEL	chr3	138664869	T	+GCAGCC	168	113	55	0.327	FOXL2	ENSG00000183770	CODON_INSERTION			1/1	YES	-	-
22	SNV	chrX	70321212	C	G	161	96	65	0.399	FOXO4	ENSG00000184481	NON_SYNONYMOUS_CODING	ENST00000374259.3:c.1132C>G	ENSP00000363377.3:p.Pro378Ala	2/3	YES	-	-
22	SNV	chrX	70321212	C	G	161	96	65	0.399	FOXO4	ENSG00000184481	NON_SYNONYMOUS_CODING	ENST00000341558.3:c.967C>G	ENSP00000342209.3:p.Pro323Ala	3/4	-	-	-
10	SNV	chr3	71096180	G	C	42	32	10	0.238	FOXP1	ENSG00000114861	NON_SYNONYMOUS_CODING	ENST00000468577.1:c.577C>G	ENSP00000418883.1:p.Gln193Glu	5/14	-	-	-
10	SNV	chr3	71096180	G	C	42	32	10	0.238	FOXP1	ENSG00000114861	NON_SYNONYMOUS_CODING	ENST00000484350.1:c.349C>G	ENSP00000417857.1:p.Gln117Glu	5/16	-	-	-
10	SNV	chr3	71096180	G	C	42	32	10	0.238	FOXP1	ENSG00000114861	NON_SYNONYMOUS_CODING	ENST00000491238.1:c.583C>G	ENSP00000420736.1:p.Gln195Glu	5/16	YES	-	-
10	SNV	chr3	71096180	G	C	42	32	10	0.238	FOXP1	ENSG00000114861	NON_SYNONYMOUS_CODING	ENST00000485326.2:c.277C>G	ENSP00000417941.2:p.Gln93Glu	4/6	-	-	-
10	SNV	chr3	71096180	G	C	42	32	10	0.238	FOXP1	ENSG00000114861	NON_SYNONYMOUS_CODING	ENST00000498215.1:c.577C>G	ENSP00000418102.1:p.Gln193Glu	6/17	-	-	-
10	SNV	chr3	71096180	G	C	42	32	10	0.238	FOXP1	ENSG00000114861	NON_SYNONYMOUS_CODING	ENST00000497355.1:c.265C>G	ENSP00000418225.1:p.Gln89Glu	3/14	-	-	-
10	SNV	chr3	71096180	G	C	42	32	10	0.238	FOXP1	ENSG00000114861	NON_SYNONYMOUS_CODING	ENST00000497553.1:c.277C>G	ENSP00000418784.1:p.Gln93Glu	4/4	-	-	-
10	SNV	chr3	71096180	G	C	42	32	10	0.238	FOXP1	ENSG00000114861	NON_SYNONYMOUS_CODING	ENST00000318789.4:c.577C>G	ENSP00000318902.4:p.Gln193Glu	10/21	-	-	-
10	SNV	chr3	71096180	G	C	42	32	10	0.238	FOXP1	ENSG00000114861	NON_SYNONYMOUS_CODING	ENST00000475937.1:c.577C>G	ENSP00000419393.1:p.Gln193Glu	9/20	-	-	-
10	SNV	chr3	71096180	G	C	42	32	10	0.238	FOXP1	ENSG00000114861	NON_SYNONYMOUS_CODING	ENST00000493089.1:c.577C>G	ENSP00000418524.1:p.Gln193Glu	10/21	-	-	-
20	SNV	chr3	71247125	G	T	308	162	146	0.472	FOXP1	ENSG00000114861	NON_SYNONYMOUS_CODING	ENST00000318721.3:c.275C>A	ENSP00000318721.3:p.Pro92His	7/7	-	-	-
S27	SNV	chr3	71050208	T	A	274	184	90	0.328	FOXP1	ENSG00000114861	SPLICE_SITE_REGION	ENST00000491238.1:c.983A>T	ENSP00000420736.1:p.His328Leu	8/16	YES	-	-
S27	SNV	chr3	71050208	T	A	274	184	90	0.328	FOXP1	ENSG00000114861	NON_SYNONYMOUS_CODING	ENST00000493089.1:c.977A>T	ENSP00000418524.1:p.His326Leu	13/21	-	-	-
S27	SNV	chr3	71050208	T	A	274	184	90	0.328	FOXP1	ENSG00000114861	NON_SYNONYMOUS_CODING	ENST00000491238.1:c.983A>T	ENSP00000420736.1:p.His328Leu	8/16	YES	-	-
S27	SNV	chr3	71050208	T	A	274	184	90	0.328	FOXP1	ENSG00000114861	SPLICE_SITE_REGION	ENST00000484350.1:c.749A>T	ENSP00000417857.1:p.His250Leu	8/16	-	-	-
S27	SNV	chr3	71050208	T	A	274	184	90	0.328	FOXP1	ENSG00000114861	SPLICE_SITE_REGION	ENST00000475937.1:c.977A>T	ENSP00000419393.1:p.His326Leu	12/20	-	-	-
S27	SNV	chr3	71050208	T	A	274	184	90	0.328	FOXP1	ENSG00000114861	NON_SYNONYMOUS_CODING	ENST00000497355.1:c.665A>T	ENSP00000418225.1:p.His222Leu	6/14	-	-	-
S27	SNV	chr3	71050208	T	A	274	184	90	0.328	FOXP1	ENSG00000114861	SPLICE_SITE_REGION	ENST00000497355.1:c.665A>T	ENSP00000418225.1:p.His222Leu	6/14	-	-	-
S27	SNV	chr3	71050208	T	A	274	184	90	0.328	FOXP1	ENSG00000114861	SPLICE_SITE_REGION	ENST00000493089.1:c.977A>T	ENSP00000418524.1:p.His326Leu	13/21	-	-	-
S27	SNV	chr3	71050208	T	A	274	184	90	0.328	FOXP1	ENSG00000114861	NON_SYNONYMOUS_CODING	ENST00000318789.4:c.977A>T	ENSP00000318902.4:p.His326Leu	13/21	-	-	-
S27	SNV	chr3	71050208	T	A	274	184	90	0.328	FOXP1	ENSG00000114861	SPLICE_SITE_REGION	ENST00000468577.1:c.977A>T	ENSP00000418883.1:p.His326Leu	8/14	-	-	-
S27	SNV	chr3	71050208	T	A	274	184	90	0.328	FOXP1	ENSG00000114861	NON_SYNONYMOUS_CODING	ENST00000468577.1:c.977A>T	ENSP00000418883.1:p.His326Leu	8/14	-	-	-
S27	SNV	chr3	71050208	T	A	274	184	90	0.328	FOXP1	ENSG00000114861	SPLICE_SITE_REGION	ENST00000498215.1:c.977A>T	ENSP00000418102.1:p.His326Leu	9/17	-	-	-
S27	SNV	chr3	71050208	T	A	274	184	90	0.328	FOXP1	ENSG00000114861	NON_SYNONYMOUS_CODING	ENST00000498215.1:c.977A>T	ENSP00000418102.1:p.His326Leu	9/17	-	-	-

S27	SNV	chr3	71050208	T	A	274	184	90	0.328	FOXP1	ENSG00000114861	NON_SYNONYMOUS_CODING	ENST00000484350.1:c.749A>T	ENSP00000417857.1:p.His250Leu	8/16	-	-	-
S27	SNV	chr3	71050208	T	A	274	184	90	0.328	FOXP1	ENSG00000114861	NON_SYNONYMOUS_CODING	ENST00000475937.1:c.977A>T	ENSP00000419393.1:p.His326Leu	12/20	-	-	-
S27	SNV	chr3	71050208	T	A	274	184	90	0.328	FOXP1	ENSG00000114861	SPLICE_SITE_REGION	ENST00000318789.4:c.977A>T	ENSP00000318902.4:p.His326Leu	13/21	-	-	-
S55	SNV	chr3	71090479	C	T	694	476	218	0.314	FOXP1	ENSG00000114861	NON_SYNONYMOUS_CODING	ENST00000497355.1:c.557G>A	ENSP00000418225.1:p.Ser186Asn	4/14	-	-	-
S55	SNV	chr3	71090479	C	T	694	476	218	0.314	FOXP1	ENSG00000114861	NON_SYNONYMOUS_CODING	ENST00000485326.2:c.569G>A	ENSP00000417941.2:p.Ser190Asn	5/6	-	-	-
S55	SNV	chr3	71090479	C	T	694	476	218	0.314	FOXP1	ENSG00000114861	SPLICE_SITE_REGION	ENST00000468577.1:c.869G>A	ENSP00000418883.1:p.Ser290Asn	6/14	-	-	-
S55	SNV	chr3	71090479	C	T	694	476	218	0.314	FOXP1	ENSG00000114861	NON_SYNONYMOUS_CODING	ENST00000484350.1:c.641G>A	ENSP00000417857.1:p.Ser214Asn	6/16	-	-	-
S55	SNV	chr3	71090479	C	T	694	476	218	0.314	FOXP1	ENSG00000114861	SPLICE_SITE_REGION	ENST00000498215.1:c.869G>A	ENSP00000418102.1:p.Ser290Asn	7/17	-	-	-
S55	SNV	chr3	71090479	C	T	694	476	218	0.314	FOXP1	ENSG00000114861	NON_SYNONYMOUS_CODING	ENST00000498215.1:c.869G>A	ENSP00000418102.1:p.Ser290Asn	7/17	-	-	-
S55	SNV	chr3	71090479	C	T	694	476	218	0.314	FOXP1	ENSG00000114861	SPLICE_SITE_REGION	ENST00000491238.1:c.875G>A	ENSP00000420736.1:p.Ser292Asn	6/16	YES	-	-
S55	SNV	chr3	71090479	C	T	694	476	218	0.314	FOXP1	ENSG00000114861	SPLICE_SITE_REGION	ENST00000493089.1:c.869G>A	ENSP00000418524.1:p.Ser290Asn	11/21	-	-	-
S55	SNV	chr3	71090479	C	T	694	476	218	0.314	FOXP1	ENSG00000114861	SPLICE_SITE_REGION	ENST00000485326.2:c.569G>A	ENSP00000417941.2:p.Ser190Asn	5/6	-	-	-
S55	SNV	chr3	71090479	C	T	694	476	218	0.314	FOXP1	ENSG00000114861	NON_SYNONYMOUS_CODING	ENST00000493089.1:c.869G>A	ENSP00000418524.1:p.Ser290Asn	11/21	-	-	-
S55	SNV	chr3	71090479	C	T	694	476	218	0.314	FOXP1	ENSG00000114861	NON_SYNONYMOUS_CODING	ENST00000491238.1:c.875G>A	ENSP00000420736.1:p.Ser292Asn	6/16	YES	-	-
S55	SNV	chr3	71090479	C	T	694	476	218	0.314	FOXP1	ENSG00000114861	SPLICE_SITE_REGION	ENST00000475937.1:c.869G>A	ENSP00000419393.1:p.Ser290Asn	10/20	-	-	-
S55	SNV	chr3	71090479	C	T	694	476	218	0.314	FOXP1	ENSG00000114861	NON_SYNONYMOUS_CODING	ENST00000318789.4:c.869G>A	ENSP00000318902.4:p.Ser290Asn	11/21	-	-	-
S55	SNV	chr3	71090479	C	T	694	476	218	0.314	FOXP1	ENSG00000114861	SPLICE_SITE_REGION	ENST00000318789.4:c.869G>A	ENSP00000318902.4:p.Ser290Asn	11/21	-	-	-
S55	SNV	chr3	71090479	C	T	694	476	218	0.314	FOXP1	ENSG00000114861	NON_SYNONYMOUS_CODING	ENST00000475937.1:c.869G>A	ENSP00000419393.1:p.Ser290Asn	10/20	-	-	-
S55	SNV	chr3	71090479	C	T	694	476	218	0.314	FOXP1	ENSG00000114861	SPLICE_SITE_REGION	ENST00000484350.1:c.641G>A	ENSP00000417857.1:p.Ser214Asn	6/16	-	-	-
S55	SNV	chr3	71090479	C	T	694	476	218	0.314	FOXP1	ENSG00000114861	SPLICE_SITE_REGION	ENST00000497355.1:c.557G>A	ENSP00000418225.1:p.Ser186Asn	4/14	-	-	-
S55	SNV	chr3	71090479	C	T	694	476	218	0.314	FOXP1	ENSG00000114861	NON_SYNONYMOUS_CODING	ENST00000468577.1:c.869G>A	ENSP00000418883.1:p.Ser290Asn	6/14	-	-	-
23	SNV	chr4	48523068	A	G	160	87	73	0.456	FRYL	ENSG00000075539	UTR_5_PRIME	ENST00000264319.7:c.-127T>C	-	55/63	-	-	-
23	SNV	chr4	48523068	A	G	160	87	73	0.456	FRYL	ENSG00000075539	UTR_5_PRIME	ENST00000507873.2:c.-127T>C	-	22/30	-	-	-
24	SNV	chr4	48581115	C	A	467	254	213	0.456	FRYL	ENSG00000075539	NON_SYNONYMOUS_CODING	ENST00000503238.1:c.2403G>T	ENSP00000426064.1:p.Trp801Cys	20/61	-	-	-
24	SNV	chr4	48581115	C	A	467	254	213	0.456	FRYL	ENSG00000075539	NON_SYNONYMOUS_CODING	ENST00000358350.4:c.2403G>T	ENSP00000351113.4:p.Trp801Cys	23/64	YES	-	-
24	SNV	chr4	48581115	C	A	467	254	213	0.456	FRYL	ENSG00000075539	NON_SYNONYMOUS_CODING	ENST00000507711.1:c.2403G>T	ENSP00000421584.1:p.Trp801Cys	23/38	-	-	-
24	SNV	chr4	48581115	C	A	467	254	213	0.456	FRYL	ENSG00000075539	UTR_5_PRIME	ENST00000264319.7:c.-5410G>T	-	23/63	-	-	-
24	SNV	chr4	48581115	C	A	467	254	213	0.456	FRYL	ENSG00000075539	NON_SYNONYMOUS_CODING	ENST00000537810.1:c.2403G>T	ENSP00000441114.1:p.Trp801Cys	23/64	-	-	-
S56	SNV	chr17	9923312	G	A	1284	656	628	0.489	GAS7	ENSG00000007237	START_GAINED	ENST00000579158.1:c.-107C>T	-	1/13	-	0.0006	rs569962505
S56	SNV	chr17	9923312	G	A	1284	656	628	0.489	GAS7	ENSG00000007237	START_GAINED	ENST00000542249.1:c.-21C>T	-	1/14	-	0.0006	rs569962505
1	SNV	chr3	128200703	G	C	288	154	134	0.465	GATA2	ENSG00000179348	NON_SYNONYMOUS_CODING	ENST00000430265.2:c.1060C>G	ENSP00000400259.2:p.Pro354Ala	5/6	-	-	-
1	SNV	chr3	128200703	G	C	288	154	134	0.465	GATA2	ENSG00000179348	NON_SYNONYMOUS_CODING	ENST00000341105.2:c.1102C>G	ENSP00000345681.2:p.Pro368Ala	5/6	YES	-	-
1	SNV	chr3	128200703	G	C	288	154	134	0.465	GATA2	ENSG00000179348	NON_SYNONYMOUS_CODING	ENST00000487848.1:c.1102C>G	ENSP00000417074.1:p.Pro368Ala	6/7	-	-	-
S02	SNV	chr3	128202755	T	C	204	160	44	0.216	GATA2	ENSG00000179348	NON_SYNONYMOUS_CODING	ENST00000341105.2:c.965A>G	ENSP00000345681.2:p.Tyr322Cys	4/6	YES	-	-
S02	SNV	chr3	128202755	T	C	204	160	44	0.216	GATA2	ENSG00000179348	NON_SYNONYMOUS_CODING	ENST00000430265.2:c.965A>G	ENSP00000400259.2:p.Tyr322Cys	4/6	-	-	-
S02	SNV	chr3	128202755	T	C	204	160	44	0.216	GATA2	ENSG00000179348	NON_SYNONYMOUS_CODING	ENST00000487848.1:c.965A>G	ENSP00000417074.1:p.Tyr322Cys	5/7	-	-	-
S46	SNV	chr3	128202767	G	A	319	180	139	0.436	GATA2	ENSG00000179348	NON_SYNONYMOUS_CODING	ENST00000430265.2:c.953C>T	ENSP00000400259.2:p.Ala318Val	4/6	-	-	COSM255084,COSM249850
S46	SNV	chr3	128202767	G	A	319	180	139	0.436	GATA2	ENSG00000179348	NON_SYNONYMOUS_CODING	ENST00000341105.2:c.953C>T	ENSP00000345681.2:p.Ala318Val	4/6	YES	-	COSM255084,COSM249850
S46	SNV	chr3	128202767	G	A	319	180	139	0.436	GATA2	ENSG00000179348	NON_SYNONYMOUS_CODING	ENST00000487848.1:c.953C>T	ENSP00000417074.1:p.Ala318Val	5/7	-	-	COSM255084,COSM249850
22	SNV	chr20	57466822	A	G	64	46	18	0.281	GNAS	ENSG00000087460	NON_SYNONYMOUS_CODING	ENST00000306090.10:c.41A>G	ENSP00000304472.10:p.Asn14Ser	2/13	-	-	-
22	SNV	chr20	57466822	A	G	64	46	18	0.281	GNAS	ENSG00000087460	NON_SYNONYMOUS_CODING	ENST00000265620.7:c.41A>G	ENSP00000265620.7:p.Asn14Ser	1/12	-	-	-
22	SNV	chr20	57466822	A	G	64	46	18	0.281	GNAS	ENSG00000087460	NON_SYNONYMOUS_CODING	ENST00000371085.3:c.41A>G	ENSP00000360126.3:p.Asn14Ser	1/13	-	-	-
22	SNV	chr20	57466822	A	G	64	46	18	0.281	GNAS	ENSG00000087460	NON_SYNONYMOUS_CODING	ENST00000371095.3:c.41A>G	ENSP00000360136.3:p.Asn14Ser	1/12	-	-	-
22	SNV	chr20	57466822	A	G	64	46	18	0.281	GNAS	ENSG00000087460	NON_SYNONYMOUS_CODING	ENST00000354359.7:c.41A>G	ENSP00000346328.7:p.Asn14Ser	1/13	-	-	-
22	SNV	chr20	57466822	A	G	64	46	18	0.281	GNAS	ENSG00000087460	NON_SYNONYMOUS_CODING	ENST00000338783.6:c.8A>G	ENSP00000345971.6:p.Asn3Ser	1/3	-	-	-
22	SNV	chr20	57466822	A	G	64	46	18	0.281	GNAS	ENSG00000087460	NON_SYNONYMOUS_CODING	ENST00000371081.1:c.41A>G	ENSP00000360122.1:p.Asn14Ser	1/4	-	-	-
26	SNV	chr20	57484596	A	G	305	187	118	0.387	GNAS	ENSG00000087460	NON_SYNONYMOUS_CODING	ENST00000354359.7:c.683A>G	ENSP00000346328.7:p.Gln228Arg	9/13	-	-	rs121913494,COSM2796,COSM27888,COSM1566191,COSM1731065
26	SNV	chr20	57484596	A	G	305	187	118	0.387	GNAS	ENSG00000087460	NON_SYNONYMOUS_CODING	ENST00000371095.3:c.638A>G	ENSP00000360136.3:p.Gln213Arg	8/12	-	-	rs121913494,COSM2796,COSM27888,COSM1566191,COSM1731065
26	SNV	chr20	57484596	A	G	305	187	118	0.387	GNAS	ENSG00000087460	NON_SYNONYMOUS_CODING	ENST00000371085.3:c.680A>G	ENSP00000360126.3:p.Gln227Arg	9/13	-	-	rs121913494,COSM2796,COSM27888,COSM1566191,COSM1731065
26	SNV	chr20	57484596	A	G	305	187	118	0.387	GNAS	ENSG00000087460	NON_SYNONYMOUS_CODING	ENST00000306090.10:c.638A>G	ENSP00000304472.10:p.Gln213Arg	9/13	-	-	rs121913494,COSM2796,COSM27888,COSM1566191,COSM1731065
26	SNV	chr20	57484596	A	G	305	187	118	0.387	GNAS	ENSG00000087460	NON_SYNONYMOUS_CODING	ENST00000265620.7:c.635A>G	ENSP00000265620.7:p.Gln212Arg	8/12	-	-	rs121913494,COSM2796,COSM27888,COSM1566191,COSM1731065
26	SNV	chr20	57484596	A	G	305	187	118	0.387	GNAS	ENSG00000087460	UTR_3_PRIME	ENST00000371075.3:c.*586A>G	-	9/13	-	-	rs121913494,COSM2796,COSM27888,COSM1566191,COSM1731065
26	SNV	chr20	57484596	A	G	305	187	118	0.387	GNAS	ENSG00000087460	UTR_3_PRIME	ENST00000313949.7:c.*583A>G	-	9/13	-	-	rs121913494,COSM2796,COSM27888,COSM1566191,COSM1731065
26	SNV	chr20	57484596	A	G	305	187	118	0.387	GNAS	ENSG00000087460	NON_SYNONYMOUS_CODING	ENST00000371100.4:c.2609A>G	ENSP00000360141.3:p.Gln870Arg	9/13	YES	-	rs121913494,COSM2796,COSM27888,COSM1566191,COSM1731065
26	SNV	chr20	57484596	A	G	305	187	118	0.387	GNAS	ENSG00000087460	NON_SYNONYMOUS_CODING	ENST00000371102.4:c.2567A>G	ENSP00000360143.4:p.Gln856Arg	8/12	-	-	rs121913494,COSM2796,COSM27888,COSM1566191,COSM1731065
S17	SNV	chr20	57428952	G	A	656	358	298	0.454	GNAS	ENSG00000087460	NON_SYNONYMOUS_CODING	ENST00000371099.2:c.632G>A	ENSP00000360140.2:p.Gly211Asp	1/2	-	-	-
S17	SNV	chr20	57428952	G	A	656	358	298	0.454	GNAS	ENSG00000087460	NON_SYNONYMOUS_CODING	ENST00000371100.4:c.632G>A	ENSP00000360141.3:p.Gly211Asp	1/13	YES	-	-
S17	SNV	chr20	57428952	G	A	656	358	298	0.454	GNAS	ENSG00000087460	NON_SYNONYMOUS_CODING	ENST00000306120.3:c.442G>A	ENSP00000302237.3:p.Ala148Thr	1/1	-	-	-
S17	SNV	chr20	57428952	G	A	656	358	298	0.454	GNAS	ENSG00000087460	NON_SYNONYMOUS_CODING	ENST00000371102.4:c.632G>A	ENSP00000360143.4:p.Gly211Asp	1/12	-	-	-
S19	SNV	chr20	57415882	C	T	353	203	150	0.425	GNAS	ENSG00000087460	NON_SYNONYMOUS_CODING	ENST00000371075.3:c.721C>T	ENSP00000360115.3:p.Pro241Ser	1/13	-	-	COSM3548233
S19	SNV	chr20	57415882	C	T	353	203	150	0.425	GNAS	ENSG00000087460	NON_SYNONYMOUS_CODING	ENST00000313949.7:c.721C>T	ENSP00000323571.7:p.Pro241Ser	1/13	-	-	COSM3548233
S19	SNV	chr20	57415882	C	T	353	203	150	0.425	GNAS	ENSG00000087460	NON_SYNONYMOUS_CODING	ENST00000419658.1:c.324C>T	ENSP00000416224.1:p.Pro109Ser	1/5	-	-	COSM3548233
S19	SNV	chr20	57415882	C	T	353	203	150	0.425	GNAS	ENSG00000087460	NON_SYNONYMOUS_CODING	ENST00000371098.2:c.721C>T	ENSP00000360139.2:p.Pro241Ser	1/4	-	-	COSM3548233
S19	SNV	chr20	57415882	C	T	353	203	150	0.425	GNAS	ENSG00000087460	NON_SYNONYMOUS_CODING	ENST00000453292.1:c.484C>T	ENSP00000392000.1:p.Pro162Ser	1/3	-	-	COSM3548233
S33	SNV	chr15	63901455	C	T	122	60	62	0.508	HERC1	ENSG00000103657	NON_SYNONYMOUS_CODING	ENST00000443617.2:c.14411G>A	ENSP00000390158.2:p.Ser4804Asn	78/78	YES	-	-
S38	SNV	chr15	63921004	C	A	368	189	179	0.486	HERC1	ENSG00000103657	NON_SYNONYMOUS_CODING	ENST00000443617.2:c.12977G>T	ENSP00000390158.2:p.Gly4326Val	70/78	YES	-	-
10	SNV	chr1	114512691	G	A	184	112	72	0.391	HIPK1	ENSG00000163349	NON_SYNONYMOUS_CODING	ENST00000369554.2:c.2750G>A	ENSP00000358567.2:p.Arg917Gln	13/15	-	-	-
10	SNV	chr1	114512691	G	A	184	112	72	0.391	HIPK1	ENSG00000163349	NON_SYNONYMOUS_CODING	ENST00000406344.1:c.1703G>A	ENSP00000384960.1:p.Arg568Gln	11/13	-	-	-
10	SNV	chr1	114512691	G	A	184	112	72	0.391	HIPK1	ENSG00000163349	NON_SYNONYMOUS_CODING	ENST00000340484.4:c.1763G>A	ENSP00000340956.4:p.Arg588Gln	13/15	-	-	-
10	SNV	chr1	114512691	G	A	184	112	72	0.391	HIPK1	ENSG00000163349	NON_SYNONYMOUS_CODING	ENST00000369555.2:c.2750G>A	ENSP00000358568.1:p.Arg917Gln	13/15	-	-	-
10	SNV	chr1	114512691	G	A	184	112	72	0.391	HIPK1	ENSG00000163349	NON_SYNONYMOUS_CODING	ENST00000369553.1:c.1703G>A	ENSP00000358566.1:p.Arg568Gln	12/14	-	-	-
10	SNV	chr1	114512691	G	A	184	112	72	0.391	HIPK1	ENSG00000163349	NON_SYNONYMOUS_CODING	ENST00000369559.4:c.2885G>A	ENSP00000358572.4:p.Arg962Gln	14/15	-	-	-
10	SNV	chr1	114512691	G	A	184	112	72	0.391	HIPK1	ENSG00000163349	NON_SYNONYMOUS_CODING	ENST00000369561.4:c.2783G>A	ENSP00000358574.4:p.Arg928Gln	14/16	-	-	-
10	SNV	chr1	114512691	G	A	184	112	72	0.391	HIPK1	ENSG00000163349	NON_SYNONYMOUS_CODING	ENST00000369558.1:c.2885G>A	ENSP00000358571.1:p.Arg962Gln	14/16	YES	-	-
10	SNV	chr1	114512691	G	A	184	112	72	0.391	HIPK1	ENSG00000163349	NON_SYNONYMOUS_CODING	ENST00000426820.2:c.2885G>A	ENSP00000407442.3:p.Arg962Gln	14/16	-	-	-
12	SNV	chr12	121439134	G	A	83	47	36	0.434	HNF1A	ENSG00000135100	UTR_3_PRIME	ENST00000541395.1:c.*939G>A	-	9/9	-	-	-
12	SNV	chr12	121439934	G	A	83	47	36	0.434	HNF1A	ENSG00000135100	UTR_3_PRIME	ENST00000257555.6:c.*939G>A	-	10/10	YES	-	-
37	SNV	chr12	121416672	G	A	68	39	29	0.426	HNF1A	ENSG00000135100	NON_SYNONYMOUS_CODING	ENST00000541395.1:c.101G>A	ENSP00000443112.1:p.Gly34Glu	1/9	-	-	-
37	SNV	chr12	121416672	G	A	68	39	29	0.426	HNF1A	ENSG00000135100	NON_SYNONYMOUS_CODING	ENST00000400024.2:c.101G>A	ENSP00000476181.1:p.Gly34Glu	1/7	-	-	-
37	SNV	chr12	121416672	G	A	68	39	29	0.426	HNF1A	ENSG00000135100	NON_SYNONYMOUS_CODING	ENST00000257555.6:c.101G>A	ENSP00000257555.4:p.Gly34Glu	1/10	YES	-	-
37	SNV	chr12	121416672	G	A	68	39	29	0.426	HNF1A	ENSG00000135100	NON_SYNONYMOUS_CODING	ENST00000402929.1:c.101G>A	ENSP00000475300.1:p.Gly34Glu	1/6	-	-	-
37	SNV	chr12	121416672	G	A	68	39	29	0.426	HNF1A	ENSG00000135100	NON_SYNONYMOUS_CODING	ENST00000544413.1:c.101G>A	ENSP00000438804.1:p.Gly34Glu	1/10	-	-	-
43	SNV	chr12	121437286	G	A	235	125	110	0.468	HNF1A	ENSG00000135100	NON_SYNONYMOUS_CODING	ENST00000544413.1:c.1717G>A	ENSP00000443112.1:p.Val573Ile	8/9	-	-	-
43	SNV	chr12	121437286	G	A	235	125	110	0.468	HNF1A	ENSG00000135100	NON_SYNONYMOUS_CODING	ENST00000544413.1:c.1645G>A	ENSP00000438804.1:p.Val549Ile	9/10	-	-	-
43	SNV	chr12	121437286	G	A	235	125	110	0.468	HNF1A	ENSG00000135100	NON_SYNONYMOUS_CODING	ENST00000257555.6:c.1624G>A	ENSP00000257555.4:p.Val542Ile	9/10	YES	-	-
S18	SNV	chr2	176972154	C	T	446	231	215	0.481	HOXD11	ENSG00000128713	NON_SYNONYMOUS_CODING	ENST00000249504.5:c.71C>T	ENSP00000249504.5:p.Ala24Val	1/2	YES	-	-
S51	SNV	chr2	176957862	C	T	359	173	186	0.518	HOXD13	ENSG00000128714	NON_SYNONYMOUS_CODING	ENST00000392539.3:c.244C>T	ENSP00000376322.3:p.Arg82Cys	1/2	YES	-	-
25	INDEL	chr6	44218831	T	-TCC	149	81	68	0.456	HSP90AB1	ENSG00000096384	CODON_CHANGE_PLUS_CODON_DELETION	ENST00000371554.1:c.1005delT	ENSP00000360609.1:p.Pro336LeufsTer34	7/12	YES	-	-
25	INDEL	chr6	44218831	T	-TCC	149	81	68	0.456	HSP90AB1	ENSG00000096384	CODON_CHANGE_PLUS_CODON_DELETION	ENST00000371646.5:c.1005delT	ENSP00000360709.5:p.Pro336LeufsTer34	7/12	-	-	-
25	INDEL	chr6	44218831	T	-TCC	149	81	68	0.456	HSP90AB1	ENSG00000096384	CODON_CHANGE_PLUS_CODON_DELETION	ENST00000353801.3:c.1005delT	ENSP00000325875.3:p.Pro336LeufsTer34	7/12	-	-	-
20	SNV	chr2	209113113	G	A	414	248	166	0.401	IDH1	ENSG00000138413	NON_SYNONYMOUS_CODING	ENST00000415282.1:c.394C>T	ENSP00000391075.1:p.Arg132Cys	3/4	-	-	rs121913499,COSM2749,COSM2747,COSM28748
20	SNV	chr2	209113113	G	A	414	248	166	0.401	IDH1	ENSG00000138413	NON_SYNONYMOUS_CODING	ENST00000415913.1:c.394C>T	ENSP00000390265.1:p.Arg132Cys	4/10	YES	-	rs121913499,COSM2749,COSM2747,COSM28748
20	SNV	chr2	209113113	G	A	414	248	166	0.401	IDH1	ENSG00000138413	NON_SYNONYMOUS_CODING	ENST00000446179.1:c.394C>T	ENSP00000410513.1:p.Arg132Cys	4/10	-	-	rs121913499,COSM2749,COSM2747,COSM28748
20	SNV	chr2	209113113	G	A	414	248	166	0.401	IDH1	ENSG00000138413	NON_SYNONYMOUS_CODING	ENST00000345146.2:c.394C>T	ENSP00000260985.2:p.Arg132Cys	4/10	-	-	rs121913499,COSM2749,COSM2747,COSM28748
24	SNV	chr2	209113112	C	T	369	221	148	0.401	IDH1	ENSG00000138413	NON_SYNONYMOUS_CODING	ENST00000415913.1:c.395G>A	ENSP00000390265.1:p.Arg132His	4/10	YES	-	rs121913500,COSM2746,COSM28750
24	SNV	chr2	209113112	C	T	369	221	148	0.401	IDH1	ENSG00000138413	NON_SYNONYMOUS_CODING	ENST00000446179.1:c.395G>A	ENSP00000410513.1:p.Arg132His	4/10	-	-	rs121913500,COSM2746,COSM28750
24	SNV	chr2	209113112	C	T	369	221	148	0.401	IDH1	ENSG00000138413	NON_SYNONYMOUS_CODING	ENST00000345146.2:c.395G>A	ENSP00000260985.2:p.Arg132His	4/10	-	-	rs121913500,COSM2746,COSM28750
24	SNV	chr2	209113112	C	T	369	221	148	0.401	IDH1	ENSG00000138413	NON_SYNONYMOUS_CODING	ENST00000415282.1:c.395G>A	ENSP00000391075.1:p.Arg132His	3/4	-	-	rs121913500,COSM2746,COSM28750
31	SNV	chr2	209113113	G	A	313	175	138	0.441	IDH1	ENSG00000138413	NON_SYNONYMOUS_CODING	ENST00000446179.1:c.394C>T	ENSP00000410513.1:p.Arg132Cys	4/10	-	-	rs121913499,COSM2749,COSM2747,COSM28748

31	SNV	chr2	209113113	G	A	313	175	138	0.441	IDH1	ENSG00000138413	NON_SYNONYMOUS_CODING	ENST00000345146.2:c.394C>T	ENSP00000260985.2:p.Arg132Cys	4/10	-	-	rs121913499,COSM28749,COSM28747,COSM28748
31	SNV	chr2	209113113	G	A	313	175	138	0.441	IDH1	ENSG00000138413	NON_SYNONYMOUS_CODING	ENST00000415282.1:c.394C>T	ENSP00000391075.1:p.Arg132Cys	3/4	-	-	rs121913499,COSM28749,COSM28747,COSM28748
31	SNV	chr2	209113113	G	A	313	175	138	0.441	IDH1	ENSG00000138413	NON_SYNONYMOUS_CODING	ENST00000415913.1:c.394C>T	ENSP00000390265.1:p.Arg132Cys	4/10	YES	-	rs121913499,COSM28749,COSM28747,COSM28748
33	SNV	chr2	209113112	C	T	465	278	187	0.402	IDH1	ENSG00000138413	NON_SYNONYMOUS_CODING	ENST00000415282.1:c.395G>A	ENSP00000391075.1:p.Arg132His	3/4	-	-	rs121913500,COSM28746,COSM28750
33	SNV	chr2	209113112	C	T	465	278	187	0.402	IDH1	ENSG00000138413	NON_SYNONYMOUS_CODING	ENST00000415913.1:c.395G>A	ENSP00000390265.1:p.Arg132His	4/10	YES	-	rs121913500,COSM28746,COSM28750
33	SNV	chr2	209113112	C	T	465	278	187	0.402	IDH1	ENSG00000138413	NON_SYNONYMOUS_CODING	ENST00000446179.1:c.395G>A	ENSP00000410513.1:p.Arg132His	4/10	-	-	rs121913500,COSM28746,COSM28750
33	SNV	chr2	209113112	C	T	465	278	187	0.402	IDH1	ENSG00000138413	NON_SYNONYMOUS_CODING	ENST00000345146.2:c.395G>A	ENSP00000260985.2:p.Arg132His	4/10	-	-	rs121913500,COSM28746,COSM28750
44	SNV	chr2	209113113	G	A	261	171	90	0.345	IDH1	ENSG00000138413	NON_SYNONYMOUS_CODING	ENST00000446179.1:c.394C>T	ENSP00000410513.1:p.Arg132Cys	4/10	-	-	rs121913499,COSM28749,COSM28747,COSM28748
44	SNV	chr2	209113113	G	A	261	171	90	0.345	IDH1	ENSG00000138413	NON_SYNONYMOUS_CODING	ENST00000345146.2:c.394C>T	ENSP00000260985.2:p.Arg132Cys	4/10	-	-	rs121913499,COSM28749,COSM28747,COSM28748
44	SNV	chr2	209113113	G	A	261	171	90	0.345	IDH1	ENSG00000138413	NON_SYNONYMOUS_CODING	ENST00000415282.1:c.394C>T	ENSP00000391075.1:p.Arg132Cys	3/4	-	-	rs121913499,COSM28749,COSM28747,COSM28748
44	SNV	chr2	209113113	G	A	261	171	90	0.345	IDH1	ENSG00000138413	NON_SYNONYMOUS_CODING	ENST00000415913.1:c.394C>T	ENSP00000390265.1:p.Arg132Cys	4/10	YES	-	rs121913499,COSM28749,COSM28747,COSM28748
62	SNV	chr2	209113113	G	A	105	75	30	0.286	IDH1	ENSG00000138413	NON_SYNONYMOUS_CODING	ENST00000446179.1:c.394C>T	ENSP00000410513.1:p.Arg132Cys	4/10	-	-	rs121913499,COSM28749,COSM28747,COSM28748
62	SNV	chr2	209113113	G	A	105	75	30	0.286	IDH1	ENSG00000138413	NON_SYNONYMOUS_CODING	ENST00000415282.1:c.394C>T	ENSP00000391075.1:p.Arg132Cys	3/4	-	-	rs121913499,COSM28749,COSM28747,COSM28748
62	SNV	chr2	209113113	G	A	105	75	30	0.286	IDH1	ENSG00000138413	NON_SYNONYMOUS_CODING	ENST00000415913.1:c.394C>T	ENSP00000390265.1:p.Arg132Cys	4/10	YES	-	rs121913499,COSM28749,COSM28747,COSM28748
62	SNV	chr2	209113113	G	A	105	75	30	0.286	IDH1	ENSG00000138413	NON_SYNONYMOUS_CODING	ENST00000345146.2:c.394C>T	ENSP00000260985.2:p.Arg132Cys	4/10	-	-	rs121913499,COSM28749,COSM28747,COSM28748
63	SNV	chr2	209113113	G	A	589	318	271	0.46	IDH1	ENSG00000138413	NON_SYNONYMOUS_CODING	ENST00000446179.1:c.394C>T	ENSP00000410513.1:p.Arg132Cys	4/10	-	-	rs121913499,COSM28749,COSM28747,COSM28748
63	SNV	chr2	209113113	G	A	589	318	271	0.46	IDH1	ENSG00000138413	NON_SYNONYMOUS_CODING	ENST00000415913.1:c.394C>T	ENSP00000390265.1:p.Arg132Cys	4/10	YES	-	rs121913499,COSM28749,COSM28747,COSM28748
63	SNV	chr2	209113113	G	A	589	318	271	0.46	IDH1	ENSG00000138413	NON_SYNONYMOUS_CODING	ENST00000415282.1:c.394C>T	ENSP00000391075.1:p.Arg132Cys	3/4	-	-	rs121913499,COSM28749,COSM28747,COSM28748
63	SNV	chr2	209113113	G	A	589	318	271	0.46	IDH1	ENSG00000138413	NON_SYNONYMOUS_CODING	ENST00000345146.2:c.394C>T	ENSP00000260985.2:p.Arg132Cys	4/10	-	-	rs121913499,COSM28749,COSM28747,COSM28748
S19	SNV	chr2	209113113	G	A	662	373	289	0.437	IDH1	ENSG00000138413	NON_SYNONYMOUS_CODING	ENST00000415282.1:c.394C>T	ENSP00000391075.1:p.Arg132Cys	3/4	-	-	rs121913499,COSM28749,COSM28747,COSM28748
S19	SNV	chr2	209113113	G	A	662	373	289	0.437	IDH1	ENSG00000138413	NON_SYNONYMOUS_CODING	ENST00000415913.1:c.394C>T	ENSP00000390265.1:p.Arg132Cys	4/10	YES	-	rs121913499,COSM28749,COSM28747,COSM28748
S19	SNV	chr2	209113113	G	A	662	373	289	0.437	IDH1	ENSG00000138413	NON_SYNONYMOUS_CODING	ENST00000446179.1:c.394C>T	ENSP00000410513.1:p.Arg132Cys	4/10	-	-	rs121913499,COSM28749,COSM28747,COSM28748
S19	SNV	chr2	209113113	G	A	662	373	289	0.437	IDH1	ENSG00000138413	NON_SYNONYMOUS_CODING	ENST00000345146.2:c.394C>T	ENSP00000260985.2:p.Arg132Cys	4/10	-	-	rs121913499,COSM28749,COSM28747,COSM28748
S30	SNV	chr2	209113113	G	A	504	301	203	0.403	IDH1	ENSG00000138413	NON_SYNONYMOUS_CODING	ENST00000446179.1:c.394C>T	ENSP00000410513.1:p.Arg132Cys	4/10	-	-	rs121913499,COSM28749,COSM28747,COSM28748
S30	SNV	chr2	209113113	G	A	504	301	203	0.403	IDH1	ENSG00000138413	NON_SYNONYMOUS_CODING	ENST00000415282.1:c.394C>T	ENSP00000391075.1:p.Arg132Cys	3/4	-	-	rs121913499,COSM28749,COSM28747,COSM28748
S30	SNV	chr2	209113113	G	A	504	301	203	0.403	IDH1	ENSG00000138413	NON_SYNONYMOUS_CODING	ENST00000345146.2:c.394C>T	ENSP00000260985.2:p.Arg132Cys	4/10	-	-	rs121913499,COSM28749,COSM28747,COSM28748
S30	SNV	chr2	209113113	G	A	504	301	203	0.403	IDH1	ENSG00000138413	NON_SYNONYMOUS_CODING	ENST00000415913.1:c.394C>T	ENSP00000390265.1:p.Arg132Cys	4/10	YES	-	rs121913499,COSM28749,COSM28747,COSM28748
S39	SNV	chr2	209110064	A	G	391	210	181	0.463	IDH1	ENSG00000138413	NON_SYNONYMOUS_CODING	ENST00000446179.1:c.499T>C	ENSP00000410513.1:p.Tyr167His	5/10	-	-	-
S39	SNV	chr2	209110064	A	G	391	210	181	0.463	IDH1	ENSG00000138413	NON_SYNONYMOUS_CODING	ENST00000415913.1:c.499T>C	ENSP00000390265.1:p.Tyr167His	5/10	YES	-	-
S39	SNV	chr2	209110064	A	G	391	210	181	0.463	IDH1	ENSG00000138413	NON_SYNONYMOUS_CODING	ENST00000345146.2:c.499T>C	ENSP00000260985.2:p.Tyr167His	5/10	-	-	-
S42	SNV	chr2	209113112	C	T	601	442	159	0.265	IDH1	ENSG00000138413	NON_SYNONYMOUS_CODING	ENST00000415913.1:c.395G>A	ENSP00000390265.1:p.Arg132His	4/10	YES	-	rs121913500,COSM28746,COSM28750
S42	SNV	chr2	209113112	C	T	601	442	159	0.265	IDH1	ENSG00000138413	NON_SYNONYMOUS_CODING	ENST00000446179.1:c.395G>A	ENSP00000410513.1:p.Arg132His	4/10	-	-	rs121913500,COSM28746,COSM28750
S42	SNV	chr2	209113112	C	T	601	442	159	0.265	IDH1	ENSG00000138413	NON_SYNONYMOUS_CODING	ENST00000415282.1:c.395G>A	ENSP00000391075.1:p.Arg132His	3/4	-	-	rs121913500,COSM28746,COSM28750
S42	SNV	chr2	209113112	C	T	601	442	159	0.265	IDH1	ENSG00000138413	NON_SYNONYMOUS_CODING	ENST00000345146.2:c.395G>A	ENSP00000260985.2:p.Arg132His	4/10	-	-	rs121913500,COSM28746,COSM28750
S45	SNV	chr2	209113113	G	A	585	325	260	0.444	IDH1	ENSG00000138413	NON_SYNONYMOUS_CODING	ENST00000345146.2:c.394C>T	ENSP00000260985.2:p.Arg132Cys	4/10	-	-	rs121913499,COSM28749,COSM28747,COSM28748
S45	SNV	chr2	209113113	G	A	585	325	260	0.444	IDH1	ENSG00000138413	NON_SYNONYMOUS_CODING	ENST00000415282.1:c.394C>T	ENSP00000391075.1:p.Arg132Cys	3/4	-	-	rs121913499,COSM28749,COSM28747,COSM28748
S45	SNV	chr2	209113113	G	A	585	325	260	0.444	IDH1	ENSG00000138413	NON_SYNONYMOUS_CODING	ENST00000415913.1:c.394C>T	ENSP00000390265.1:p.Arg132Cys	4/10	YES	-	rs121913499,COSM28749,COSM28747,COSM28748
S45	SNV	chr2	209113113	G	A	585	325	260	0.444	IDH1	ENSG00000138413	NON_SYNONYMOUS_CODING	ENST00000446179.1:c.394C>T	ENSP00000410513.1:p.Arg132Cys	4/10	-	-	rs121913499,COSM28749,COSM28747,COSM28748
S52	SNV	chr2	209113113	G	A	707	419	288	0.407	IDH1	ENSG00000138413	NON_SYNONYMOUS_CODING	ENST00000415282.1:c.394C>T	ENSP00000391075.1:p.Arg132Cys	3/4	-	-	rs121913499,COSM28749,COSM28747,COSM28748
S52	SNV	chr2	209113113	G	A	707	419	288	0.407	IDH1	ENSG00000138413	NON_SYNONYMOUS_CODING	ENST00000415913.1:c.394C>T	ENSP00000390265.1:p.Arg132Cys	4/10	YES	-	rs121913499,COSM28749,COSM28747,COSM28748
S52	SNV	chr2	209113113	G	A	707	419	288	0.407	IDH1	ENSG00000138413	NON_SYNONYMOUS_CODING	ENST00000345146.2:c.394C>T	ENSP00000260985.2:p.Arg132Cys	4/10	-	-	rs121913499,COSM28749,COSM28747,COSM28748
S52	SNV	chr2	209113113	G	A	707	419	288	0.407	IDH1	ENSG00000138413	NON_SYNONYMOUS_CODING	ENST00000446179.1:c.394C>T	ENSP00000410513.1:p.Arg132Cys	4/10	-	-	rs121913499,COSM28749,COSM28747,COSM28748
S56	SNV	chr2	209113113	G	A	939	546	393	0.419	IDH1	ENSG00000138413	NON_SYNONYMOUS_CODING	ENST00000446179.1:c.394C>T	ENSP00000410513.1:p.Arg132Cys	4/10	-	-	rs121913499,COSM28749,COSM28747,COSM28748
S56	SNV	chr2	209113113	G	A	939	546	393	0.419	IDH1	ENSG00000138413	NON_SYNONYMOUS_CODING	ENST00000345146.2:c.394C>T	ENSP00000260985.2:p.Arg132Cys	4/10	-	-	rs121913499,COSM28749,COSM28747,COSM28748
S56	SNV	chr2	209113113	G	A	939	546	393	0.419	IDH1	ENSG00000138413	NON_SYNONYMOUS_CODING	ENST00000415282.1:c.394C>T	ENSP00000391075.1:p.Arg132Cys	3/4	-	-	rs121913499,COSM28749,COSM28747,COSM28748
S56	SNV	chr2	209113113	G	A	939	546	393	0.419	IDH1	ENSG00000138413	NON_SYNONYMOUS_CODING	ENST00000415913.1:c.394C>T	ENSP00000390265.1:p.Arg132Cys	4/10	YES	-	rs121913499,COSM28749,COSM28747,COSM28748
S58	SNV	chr2	209113113	G	A	795	408	387	0.486	IDH1	ENSG00000138413	NON_SYNONYMOUS_CODING	ENST00000446179.1:c.394C>T	ENSP00000410513.1:p.Arg132Cys	4/10	-	-	rs121913499,COSM28749,COSM28747,COSM28748
S58	SNV	chr2	209113113	G	A	795	408	387	0.486	IDH1	ENSG00000138413	NON_SYNONYMOUS_CODING	ENST00000345146.2:c.394C>T	ENSP00000260985.2:p.Arg132Cys	4/10	-	-	rs121913499,COSM28749,COSM28747,COSM28748
S58	SNV	chr2	209113113	G	A	795	408	387	0.486	IDH1	ENSG00000138413	NON_SYNONYMOUS_CODING	ENST00000415913.1:c.394C>T	ENSP00000390265.1:p.Arg132Cys	4/10	YES	-	rs121913499,COSM28749,COSM28747,COSM28748
S58	SNV	chr2	209113113	G	A	795	408	387	0.486	IDH1	ENSG00000138413	NON_SYNONYMOUS_CODING	ENST00000415282.1:c.394C>T	ENSP00000391075.1:p.Arg132Cys	3/4	-	-	rs121913499,COSM28749,COSM28747,COSM28748
S63	SNV	chr2	209113113	G	A	654	425	229	0.35	IDH1	ENSG00000138413	NON_SYNONYMOUS_CODING	ENST00000446179.1:c.394C>T	ENSP00000410513.1:p.Arg132Cys	4/10	-	-	rs121913499,COSM28749,COSM28747,COSM28748
S63	SNV	chr2	209113113	G	A	654	425	229	0.35	IDH1	ENSG00000138413	NON_SYNONYMOUS_CODING	ENST00000415282.1:c.394C>T	ENSP00000391075.1:p.Arg132Cys	3/4	-	-	rs121913499,COSM28749,COSM28747,COSM28748
S63	SNV	chr2	209113113	G	A	654	425	229	0.35	IDH1	ENSG00000138413	NON_SYNONYMOUS_CODING	ENST00000415913.1:c.394C>T	ENSP00000390265.1:p.Arg132Cys	4/10	YES	-	rs121913499,COSM28749,COSM28747,COSM28748
S63	SNV	chr2	209113113	G	A	654	425	229	0.35	IDH1	ENSG00000138413	NON_SYNONYMOUS_CODING	ENST00000345146.2:c.394C>T	ENSP00000260985.2:p.Arg132Cys	4/10	-	-	rs121913499,COSM28749,COSM28747,COSM28748
12	SNV	chr15	90631934	C	T	198	111	87	0.439	IDH2	ENSG00000182054	NON_SYNONYMOUS_CODING	ENST00000540499.2:c.263G>A	ENSP00000446147.2:p.Arg88Gln	4/11	-	-	rs121913502,COSM41875,COSM41590,CM106818
12	SNV	chr15	90631934	C	T	198	111	87	0.439	IDH2	ENSG00000182054	NON_SYNONYMOUS_CODING	ENST00000330062.3:c.419G>A	ENSP00000331897.3:p.Arg140Gln	4/11	YES	-	rs121913502,COSM41875,COSM41590,CM106818
12	SNV	chr15	90631934	C	T	198	111	87	0.439	IDH2	ENSG00000182054	NON_SYNONYMOUS_CODING	ENST00000539790.1:c.29G>A	ENSP00000438457.1:p.Arg10Gln	2/9	-	-	rs121913502,COSM41875,COSM41590,CM106818
36	SNV	chr15	90631934	C	T	259	130	129	0.498	IDH2	ENSG00000182054	NON_SYNONYMOUS_CODING	ENST00000539790.1:c.29G>A	ENSP00000438457.1:p.Arg10Gln	2/9	-	-	rs121913502,COSM41875,COSM41590,CM106818
36	SNV	chr15	90631934	C	T	259	130	129	0.498	IDH2	ENSG00000182054	NON_SYNONYMOUS_CODING	ENST00000330062.3:c.419G>A	ENSP00000331897.3:p.Arg140Gln	4/11	YES	-	rs121913502,COSM41875,COSM41590,CM106818
36	SNV	chr15	90631934	C	T	259	130	129	0.498	IDH2	ENSG00000182054	NON_SYNONYMOUS_CODING	ENST00000540499.2:c.263G>A	ENSP00000446147.2:p.Arg88Gln	4/11	-	-	rs121913502,COSM41875,COSM41590,CM106818
40	SNV	chr15	90631838	C	T	123	72	51	0.415	IDH2	ENSG00000182054	NON_SYNONYMOUS_CODING	ENST00000539790.1:c.125G>A	ENSP00000438457.1:p.Arg42Lys	2/9	-	-	rs121913503,COSM33732,COSM33733,COSM1636997
40	SNV	chr15	90631838	C	T	123	72	51	0.415	IDH2	ENSG00000182054	NON_SYNONYMOUS_CODING	ENST00000330062.3:c.515G>A	ENSP00000331897.3:p.Arg172Lys	4/11	YES	-	rs121913503,COSM33732,COSM33733,COSM1636997
40	SNV	chr15	90631838	C	T	123	72	51	0.415	IDH2	ENSG00000182054	NON_SYNONYMOUS_CODING	ENST00000540499.2:c.359G>A	ENSP00000446147.2:p.Arg120Lys	4/11	-	-	rs121913503,COSM33732,COSM33733,COSM1636997
58	SNV	chr15	90631835	T	G	226	137	89	0.394	IDH2	ENSG00000182054	NON_SYNONYMOUS_CODING	ENST00000540499.2:c.362A>C	ENSP00000446147.2:p.His121Pro	4/11	-	-	COSM3999681
58	SNV	chr15	90631836	G	C	260	169	91	0.35	IDH2	ENSG00000182054	NON_SYNONYMOUS_CODING	ENST00000540499.2:c.361C>G	ENSP00000446147.2:p.His121Asp	4/11	-	-	COSM3999682
58	SNV	chr15	90631836	G	C	260	169	91	0.35	IDH2	ENSG00000182054	NON_SYNONYMOUS_CODING	ENST00000539790.1:c.127C>G	ENSP00000438457.1:p.His43Asp	2/9	-	-	COSM3999682
58	SNV	chr15	90631836	G	C	260	169	91	0.35	IDH2	ENSG00000182054	NON_SYNONYMOUS_CODING	ENST00000330062.3:c.517C>G	ENSP00000331897.3:p.His173Asp	4/11	YES	-	COSM3999682
58	SNV	chr15	90631837	C	A	227	139	88	0.342	IDH2	ENSG00000182054	NON_SYNONYMOUS_CODING	ENST00000539790.1:c.126G>T	ENSP00000438457.1:p.Arg42Ser	2/9	-	-	COSM34090,COSM133672
58	SNV	chr15	90631837	C	A	227	139	88	0.342	IDH2	ENSG00000182054	NON_SYNONYMOUS_CODING	ENST00000540499.2:c.360G>T	ENSP00000446147.2:p.Arg120Ser	4/11	-	-	COSM34090,COSM133672
58	SNV	chr15	90631837	C	A	227	139	88	0.342	IDH2	ENSG00000182054	NON_SYNONYMOUS_CODING	ENST00000330062.3:c.516G>T	ENSP00000331897.3:p.Arg172Ser	4/11	YES	-	COSM34090,COSM133672
58	SNV	chr15	90631835	T	G	226	137	89	0.394	IDH2	ENSG00000182054	NON_SYNONYMOUS_CODING	ENST00000539790.1:c.128A>C	ENSP00000438457.1:p.His43Pro	2/9	-	-	COSM3999681
58	SNV	chr15	90631835	T	G	226	137	89	0.394	IDH2	ENSG00000182054	NON_SYNONYMOUS_CODING	ENST00000330062.3:c.518A>C	ENSP00000331897.3:p.His173Pro	4/11	YES	-	COSM3999681
S06	SNV	chr15	90631934	C	T	298	154	144	0.483	IDH2	ENSG00000182054	NON_SYNONYMOUS_CODING	ENST00000330062.3:c.419G>A	ENSP00000331897.3:p.Arg140Gln	4/11	YES	-	rs121913502,COSM41875,COSM41590,CM106818
S06	SNV	chr15	90631934	C	T	298	154	144	0.483	IDH2	ENSG00000182054	NON_SYNONYMOUS_CODING	ENST00000539790.1:c.29G>A	ENSP00000438457.1:p.Arg10Gln	2/9	-	-	rs121913502,COSM41875,COSM41590,CM106818
S06	SNV	chr15	90631934	C	T	298	154	144	0.483	IDH2	ENSG00000182054	NON_SYNONYMOUS_CODING	ENST00000540499.2:c.263G>A	ENSP00000446147.2:p.Arg88Gln	4/11	-	-	rs121913502,COSM41875,COSM41590,CM106818
S14	SNV	chr15	90631934	C	T	200	109	91	0.455	IDH2	ENSG00000182054	NON_SYNONYMOUS_CODING	ENST00000330062.3:c.419G>A	ENSP00000331897.3:p.Arg140Gln	4/11	YES	-	rs121913502,COSM41875,COSM41590,CM106818
S14	SNV	chr15	90631934	C	T	200	109	91	0.455	IDH2	ENSG00000182054	NON_SYNONYMOUS_CODING	ENST00000540499.2:c.263G>A	ENSP00000446147.2:p.Arg88Gln	4/11	-	-	rs121913502,COSM41875,COSM41590,CM106818
S14	SNV	chr15	90631934	C	T	200	109	91	0.455	IDH2	ENSG00000182054	NON_SYNONYMOUS_CODING	ENST00000539790.1:c.29G>A	ENSP00000438457.1:p.Arg10Gln	2/9	-	-	rs121913502,COSM41875,COSM41590,CM106818
S18	SNV	chr15	90631934	C	T	405	205	200	0.494	IDH2	ENSG00000182054	NON_SYNONYMOUS_CODING	ENST00000540499.2:c.263G>A	ENSP00000446147.2:p.Arg88Gln	4/11	-	-	rs121913502,COSM41875,COSM41590,CM106818
S18	SNV	chr15	90631934	C	T	405	205	200	0.494	IDH2	ENSG00000182054	NON_SYNONYMOUS_CODING	ENST00000539790.1:c.29G>A	ENSP00000438457.1:p.Arg10Gln	2/9	-	-	rs121913502,COSM41875,COSM41590,CM106818
S18	SNV	chr15	90631934	C	T	405	205	200	0.494	IDH2	ENSG00000182054	NON_SYNONYMOUS_CODING	ENST00000330062.3:c.419G>A	ENSP00000331897.3:p.Arg140Gln	4/11	YES	-	rs121913502,COSM41875,COSM41590,CM106818
S35	SNV	chr15	90631934	C	T	389	210	179	0.46	IDH2	ENSG00000182054	NON_SYNONYMOUS_CODING	ENST00000539790.1:c.29G>A	ENSP00000438457.1:p.Arg10Gln	2/9	-	-	rs121913502,COSM41875,COSM41590,CM106818
S35	SNV	chr15	90631934	C	T	389	210	179	0.46	IDH2	ENSG00000182054	NON_SYNONYMOUS_CODING	ENST00000540499.2:c.263G>A	ENSP00000446147.2:p.Arg88Gln	4/11	-	-	rs121913502,COSM41875,COSM41590,CM106818
S35	SNV	chr15	90631934	C	T	389	210	179	0.46	IDH2	ENSG00000182054	NON_SYNONYMOUS_CODING	ENST00000330062.3:c.419G>A	ENSP00000331897.3:p.Arg140Gln	4/11	YES	-	rs121913502,COSM41875,COSM41590,CM106818
S36	SNV	chr15	90631934	C	T	507	277	230	0.454	IDH2	ENSG00000182054	NON_SYNONYMOUS_CODING	ENST00000540499.2:c.263G>A	ENSP00000446147.2:p.Arg88Gln	4/11	-	-	rs121913502,COSM41875,COSM41590,CM106818
S36	SNV	chr15	90631934	C	T	507	277	230	0.454	IDH2	ENSG00000182054	NON_SYNONYMOUS_CODING	ENST00000539790.1:c.29G>A	ENSP00000438457.1:p.Arg10Gln	2/9	-	-	rs121913502,COSM41875,COSM41590,CM106818
S36	SNV	chr15	90631934	C	T	507	277	230	0.454	IDH2	ENSG00000182054	NON_SYNONYMOUS_CODING	ENST00000330062.3:c.419G>A	ENSP00000331897.3:p.Arg140Gln	4/11	YES	-	rs121913502,COSM41875,COSM41590,CM106818
S53	SNV	chr15	90631934	C	T	413	252	161	0.39	IDH2	ENSG00000182054	NON_SYNONYMOUS_CODING	ENST00000540499.2:c.263G>A	ENSP00000446147.2:p.Arg88Gln	4/11	-	-	rs121913502,COSM41875,COSM41590,CM106818
S53	SNV	chr15	90631934	C	T	413	252	161	0.39	IDH2	ENSG00000182054	NON_SYNONYMOUS_CODING	ENST00000330062.3:c.419G>A	ENSP00000331897.3:p.Arg140Gln	4/11	YES	-	rs121913502,COSM41875,COSM41590,CM106818
S53	SNV	chr15	90631934	C	T	413	252	161	0.39	IDH2	ENSG00000182054	NON_SYNONYMOUS_CODING	ENST00000539790.1:c.29G>A	ENSP00000438457.1:p.Arg10Gln	2/9	-	-	rs121913502,COSM41875,COSM41590,CM106818
S48	SNV	chr15	99500419	G	T	671	326	345	0.514	IGF1R	ENSG00000140443	NON_SYNONYMOUS_CODING	ENST00000288035.6:c.3852G>T	ENSP00000288035.6:p.Glu1284Asp	21/21	YES	-	-
S48	SNV	chr15	99500419	G	T	671	326	345	0.514	IGF1R	ENSG00000140443	NON_SYNONYMOUS_CODING	ENST00000558762.1:c.3849G>T	ENSP00000453007.1:p.Glu1283Asp	21/21	-	-	-
S29	INDEL	chr7	50450369	A	+GG	215	133	82	0.376	IKZF1	ENSG00000185811	FRAME_SHIFT	-	-	4/7	-	-	-
S29	INDEL	chr7	50450369	A	+GG	215	133	82	0.376	IKZF1	ENSG00000185811	FRAME_SHIFT	-	-	5/7	YES	-	-
S29	INDEL	chr7	50450369	A	+GG	215	133	82	0.376	IKZF1	ENSG00000185811	FRAME_SHIFT	-	-	4/7	-	-	-
S29	INDEL	chr7	50450369	A	+GG	215	133	82	0.376	IKZF1	ENSG00000185811	FRAME_SHIFT	-	-	5/8	-	-	-
S29	INDEL	chr7	50450369	A	+GG	215	133	82	0.376	IKZF1	ENSG00000185811	FRAME_SHIFT	-	-	5/6	-	-	-
S29	INDEL	chr7	50450369	A	+GG	215	133	82	0.376	IKZF1	ENSG00000185811	FRAME_SHIFT	-	-	5/7	-	-	-
S29	INDEL	chr7	50450369	A	+GG	215	133	82	0.376	IKZF1	ENSG00000185811	FRAME_SHIFT	-	-	5/8	-	-	-

S54	SNV	chr7	50450288	G	A	642	336	306	0.477	IKZF1	ENSG00000185811	NON_SYNONYMOUS_CODING	ENST00000359197.5:c.472G>A	ENSP00000352123.5:p.Gly158Ser	5/7	-	-	COSM249841,COSM303844
S54	SNV	chr7	50450288	G	A	642	336	306	0.477	IKZF1	ENSG00000185811	NON_SYNONYMOUS_CODING	ENST00000439701.1:c.472G>A	ENSP00000413025.1:p.Gly158Ser	5/7	YES	-	COSM249841,COSM303844
S54	SNV	chr7	50450288	G	A	642	336	306	0.477	IKZF1	ENSG00000185811	NON_SYNONYMOUS_CODING	ENST00000357364.4:c.472G>A	ENSP00000349928.4:p.Gly158Ser	5/6	-	-	COSM249841,COSM303844
S54	SNV	chr7	50450288	G	A	642	336	306	0.477	IKZF1	ENSG00000185811	NON_SYNONYMOUS_CODING	ENST00000440768.2:c.472G>A	ENSP00000401507.2:p.Gly158Ser	5/8	-	-	COSM249841,COSM303844
S54	SNV	chr7	50450288	G	A	642	336	306	0.477	IKZF1	ENSG00000185811	NON_SYNONYMOUS_CODING	ENST00000331340.3:c.472G>A	ENSP00000331614.3:p.Gly158Ser	5/8	-	-	COSM249841,COSM303844
S54	SNV	chr7	50450288	G	A	642	336	306	0.477	IKZF1	ENSG00000185811	NON_SYNONYMOUS_CODING	ENST00000343574.5:c.211G>A	ENSP00000342750.5:p.Gly71Ser	4/7	-	-	COSM249841,COSM303844
S54	SNV	chr7	50450288	G	A	642	336	306	0.477	IKZF1	ENSG00000185811	NON_SYNONYMOUS_CODING	ENST00000438033.1:c.211G>A	ENSP00000396554.1:p.Gly71Ser	4/7	-	-	COSM249841,COSM303844
S58	SNV	chr7	50367257	G	A	911	443	468	0.514	IKZF1	ENSG00000185811	NON_SYNONYMOUS_CODING	ENST00000343574.5:c.64G>A	ENSP00000342750.5:p.Asp22Asn	3/7	-	-	COSM3265388
S58	SNV	chr7	50367257	G	A	911	443	468	0.514	IKZF1	ENSG00000185811	NON_SYNONYMOUS_CODING	ENST00000440768.2:c.64G>A	ENSP00000401507.2:p.Asp22Asn	3/8	-	-	COSM3265388
S58	SNV	chr7	50367257	G	A	911	443	468	0.514	IKZF1	ENSG00000185811	NON_SYNONYMOUS_CODING	ENST00000357364.4:c.64G>A	ENSP00000349928.4:p.Asp22Asn	3/6	-	-	COSM3265388
S58	SNV	chr7	50367257	G	A	911	443	468	0.514	IKZF1	ENSG00000185811	NON_SYNONYMOUS_CODING	ENST00000331340.3:c.64G>A	ENSP00000331614.3:p.Asp22Asn	3/8	-	-	COSM3265388
S58	SNV	chr7	50367257	G	A	911	443	468	0.514	IKZF1	ENSG00000185811	NON_SYNONYMOUS_CODING	ENST00000346667.4:c.64G>A	ENSP00000340080.4:p.Asp22Asn	3/4	-	-	COSM3265388
S58	SNV	chr7	50367257	G	A	911	443	468	0.514	IKZF1	ENSG00000185811	NON_SYNONYMOUS_CODING	ENST00000349824.4:c.64G>A	ENSP00000342485.4:p.Asp22Asn	3/5	-	-	COSM3265388
S58	SNV	chr7	50367257	G	A	911	443	468	0.514	IKZF1	ENSG00000185811	NON_SYNONYMOUS_CODING	ENST00000359197.5:c.64G>A	ENSP00000352123.5:p.Asp22Asn	3/7	-	-	COSM3265388
S58	SNV	chr7	50367257	G	A	911	443	468	0.514	IKZF1	ENSG00000185811	NON_SYNONYMOUS_CODING	ENST00000439701.1:c.64G>A	ENSP00000413025.1:p.Asp22Asn	3/7	YES	-	COSM3265388
S58	SNV	chr7	50367257	G	A	911	443	468	0.514	IKZF1	ENSG00000185811	NON_SYNONYMOUS_CODING	ENST00000413698.1:c.64G>A	ENSP00000388478.1:p.Asp22Asn	3/4	-	-	COSM3265388
S58	SNV	chr7	50367257	G	A	911	443	468	0.514	IKZF1	ENSG00000185811	NON_SYNONYMOUS_CODING	ENST00000426121.1:c.13G>A	ENSP00000409588.1:p.Asp5Asn	1/2	-	-	COSM3265388
S58	SNV	chr7	50367257	G	A	911	443	468	0.514	IKZF1	ENSG00000185811	NON_SYNONYMOUS_CODING	ENST00000438033.1:c.64G>A	ENSP00000396554.1:p.Asp22Asn	3/7	-	-	COSM3265388
34	SNV	chr16	27460524	G	A	234	108	126	0.538	IL21R	ENSG00000103522	NON_SYNONYMOUS_CODING	ENST00000395754.4:c.1537G>A	ENSP00000379103.4:p.Glu513Lys	9/9	-	-	COSM255978
34	SNV	chr16	27460524	G	A	234	108	126	0.538	IL21R	ENSG00000103522	NON_SYNONYMOUS_CODING	ENST00000395755.1:c.1537G>A	ENSP00000379104.1:p.Glu513Lys	9/9	-	-	COSM255978
34	SNV	chr16	27460524	G	A	234	108	126	0.538	IL21R	ENSG00000103522	NON_SYNONYMOUS_CODING	ENST00000337929.3:c.1537G>A	ENSP00000338010.3:p.Glu513Lys	9/9	YES	-	COSM255978
47	SNV	chr5	55251969	G	T	143	74	69	0.482	IL6ST	ENSG00000134352	NON_SYNONYMOUS_CODING	ENST00000336909.5:c.1151C>A	ENSP00000338799.5:p.Ala384Asp	8/15	-	-	-
47	SNV	chr5	55251969	G	T	143	74	69	0.482	IL6ST	ENSG00000134352	UTR_3_PRIME	ENST00000536319.1:c.*78C>A	-	7/13	-	-	-
47	SNV	chr5	55251969	G	T	143	74	69	0.482	IL6ST	ENSG00000134352	NON_SYNONYMOUS_CODING	ENST00000381298.2:c.1151C>A	ENSP00000370698.2:p.Ala384Asp	10/17	YES	-	-
47	SNV	chr5	55251969	G	T	143	74	69	0.482	IL6ST	ENSG00000134352	UTR_3_PRIME	ENST00000381287.4:c.*78C>A	-	7/14	-	-	-
47	SNV	chr5	55251969	G	T	143	74	69	0.482	IL6ST	ENSG00000134352	UTR_3_PRIME	ENST00000522633.2:c.*78C>A	-	8/13	-	-	-
47	SNV	chr5	55251969	G	T	143	74	69	0.482	IL6ST	ENSG00000134352	NON_SYNONYMOUS_CODING	ENST00000381294.3:c.1151C>A	ENSP00000370694.3:p.Ala384Asp	8/14	-	-	-
47	SNV	chr5	55251969	G	T	143	74	69	0.482	IL6ST	ENSG00000134352	NON_SYNONYMOUS_CODING	ENST00000502326.3:c.1151C>A	ENSP00000462158.1:p.Ala384Asp	9/16	-	-	-
S27	SNV	chr13	111371893	C	T	638	324	314	0.492	ING1	ENSG00000153487	NON_SYNONYMOUS_CODING	ENST00000375774.3:c.883C>T	ENSP00000364929.3:p.Arg295Trp	2/2	YES	-	-
S27	SNV	chr13	111371893	C	T	638	324	314	0.492	ING1	ENSG00000153487	NON_SYNONYMOUS_CODING	ENST00000375775.3:c.247C>T	ENSP00000364930.3:p.Arg83Trp	2/2	-	-	-
S27	SNV	chr13	111371893	C	T	638	324	314	0.492	ING1	ENSG00000153487	NON_SYNONYMOUS_CODING	ENST00000338450.7:c.322C>T	ENSP00000345202.7:p.Arg108Trp	2/2	-	-	-
S27	SNV	chr13	111371893	C	T	638	324	314	0.492	ING1	ENSG00000153487	NON_SYNONYMOUS_CODING	ENST00000333219.7:c.454C>T	ENSP00000328436.7:p.Arg152Trp	2/2	-	-	-
S65	SNV	chr6	4005097	C	G	742	380	362	0.488	IRF4	ENSG00000137265	NON_SYNONYMOUS_CODING	ENST00000380956.4:c.1119C>G	ENSP00000370348.1:p.His373Gln	8/9	YES	-	-
47	SNV	chr5	156671451	C	T	118	59	59	0.5	ITK	ENSG00000113263	NON_SYNONYMOUS_CODING	ENST00000422843.3:c.1412C>T	ENSP00000398655.3:p.Ala471Val	13/17	YES	-	-
S52	SNV	chr1	65301857	G	A	621	317	304	0.49	JAK1	ENSG00000162434	NON_SYNONYMOUS_CODING	ENST00000342505.4:c.3182C>T	ENSP00000343204.4:p.Ala1061Val	23/25	YES	-	-
S30	SNV	chr9	5073770	G	T	476	40	436	0.916	JAK2	ENSG00000096968	NON_SYNONYMOUS_CODING	ENST00000539801.1:c.1849G>T	ENSP00000440387.1:p.Val617Phe	13/24	-	-	rs77375493,COSM12600,COSM29117
S30	SNV	chr9	5073770	G	T	476	40	436	0.916	JAK2	ENSG00000096968	NON_SYNONYMOUS_CODING	ENST00000544510.1:c.1402G>T	ENSP00000443103.1:p.Val468Phe	11/22	-	-	rs77375493,COSM12600,COSM29117
S30	SNV	chr9	5073770	G	T	476	40	436	0.916	JAK2	ENSG00000096968	NON_SYNONYMOUS_CODING	ENST00000381652.3:c.1849G>T	ENSP00000371067.3:p.Val617Phe	14/25	YES	-	rs77375493,COSM12600,COSM29117
S51	SNV	chr9	5073770	G	T	494	318	176	0.356	JAK2	ENSG00000096968	NON_SYNONYMOUS_CODING	ENST00000381652.3:c.1849G>T	ENSP00000371067.3:p.Val617Phe	14/25	YES	-	rs77375493,COSM12600,COSM29117
S51	SNV	chr9	5073770	G	T	494	318	176	0.356	JAK2	ENSG00000096968	NON_SYNONYMOUS_CODING	ENST00000539801.1:c.1849G>T	ENSP00000440387.1:p.Val617Phe	13/24	-	-	rs77375493,COSM12600,COSM29117
S51	SNV	chr9	5073770	G	T	494	318	176	0.356	JAK2	ENSG00000096968	NON_SYNONYMOUS_CODING	ENST00000544510.1:c.1402G>T	ENSP00000443103.1:p.Val468Phe	11/22	-	-	rs77375493,COSM12600,COSM29117
12	SNV	chr19	17953950	G	C	174	111	63	0.362	JAK3	ENSG00000105639	NON_SYNONYMOUS_CODING	ENST00000534444.1:c.452C>G	ENSP00000436421.1:p.Pro151Arg	5/23	-	0.0018	rs55778349,JAK3base_D0095:g.6333C>G,COSM35867,COSM3530608,COSM3530609,CM000171
12	SNV	chr19	17953950	G	C	174	111	63	0.362	JAK3	ENSG00000105639	NON_SYNONYMOUS_CODING	ENST00000458235.1:c.452C>G	ENSP00000391676.1:p.Pro151Arg	5/24	YES	0.0018	rs55778349,JAK3base_D0095:g.6333C>G,COSM35867,COSM3530608,COSM3530609,CM000171
12	SNV	chr19	17953950	G	C	174	111	63	0.362	JAK3	ENSG00000105639	NON_SYNONYMOUS_CODING	ENST00000527670.1:c.452C>G	ENSP00000432511.1:p.Pro151Arg	4/23	-	0.0018	rs55778349,JAK3base_D0095:g.6333C>G,COSM35867,COSM3530608,COSM3530609,CM000171
38	SNV	chr19	17953950	G	C	267	141	126	0.472	JAK3	ENSG00000105639	NON_SYNONYMOUS_CODING	ENST00000534444.1:c.452C>G	ENSP00000436421.1:p.Pro151Arg	5/23	-	0.0018	rs55778349,JAK3base_D0095:g.6333C>G,COSM35867,COSM3530608,COSM3530609,CM000171
38	SNV	chr19	17953950	G	C	267	141	126	0.472	JAK3	ENSG00000105639	NON_SYNONYMOUS_CODING	ENST00000458235.1:c.452C>G	ENSP00000391676.1:p.Pro151Arg	5/24	YES	0.0018	rs55778349,JAK3base_D0095:g.6333C>G,COSM35867,COSM3530608,COSM3530609,CM000171
38	SNV	chr19	17953950	G	C	267	141	126	0.472	JAK3	ENSG00000105639	NON_SYNONYMOUS_CODING	ENST00000527670.1:c.452C>G	ENSP00000432511.1:p.Pro151Arg	4/23	-	0.0018	rs55778349,JAK3base_D0095:g.6333C>G,COSM35867,COSM3530608,COSM3530609,CM000171
S66	SNV	chr19	17942208	C	G	349	174	175	0.501	JAK3	ENSG00000105639	SPLICE_SITE_REGION	ENST00000534444.1:c.2807G>C	ENSP00000436421.1:p.Gly936Ala	21/23	-	-	-
S66	SNV	chr19	17942208	C	G	349	174	175	0.501	JAK3	ENSG00000105639	SPLICE_SITE_REGION	ENST00000458235.1:c.2807G>C	ENSP00000391676.1:p.Gly936Ala	21/24	YES	-	-
S66	SNV	chr19	17942208	C	G	349	174	175	0.501	JAK3	ENSG00000105639	SPLICE_SITE_REGION	ENST00000527670.1:c.2807G>C	ENSP00000432511.1:p.Gly936Ala	20/23	-	-	-
S66	SNV	chr19	17942208	C	G	349	174	175	0.501	JAK3	ENSG00000105639	NON_SYNONYMOUS_CODING	ENST00000458235.1:c.2807G>C	ENSP00000391676.1:p.Gly936Ala	21/24	YES	-	-
S66	SNV	chr19	17942208	C	G	349	174	175	0.501	JAK3	ENSG00000105639	NON_SYNONYMOUS_CODING	ENST00000527670.1:c.2807G>C	ENSP00000432511.1:p.Gly936Ala	20/23	-	-	-
S66	SNV	chr19	17942208	C	G	349	174	175	0.501	JAK3	ENSG00000105639	NON_SYNONYMOUS_CODING	ENST00000534444.1:c.2807G>C	ENSP00000436421.1:p.Gly936Ala	21/23	-	-	-
10	SNV	chr6	15487765	C	G	158	106	52	0.329	JARID2	ENSG00000008083	NON_SYNONYMOUS_CODING	ENST00000341776.2:c.898C>G	ENSP00000341280.2:p.Arg300Gly	6/18	YES	-	rs375479047,COSM171612
10	SNV	chr6	15487765	C	G	158	106	52	0.329	JARID2	ENSG00000008083	NON_SYNONYMOUS_CODING	ENST00000541660.1:c.784C>G	ENSP00000444623.1:p.Arg262Gly	5/12	-	-	rs375479047,COSM171612
10	SNV	chr6	15487765	C	G	158	106	52	0.329	JARID2	ENSG00000008083	NON_SYNONYMOUS_CODING	ENST00000397311.3:c.382C>G	ENSP00000380478.3:p.Arg128Gly	6/18	-	-	rs375479047,COSM171612
31	SNV	chr6	15517413	G	A	527	287	240	0.455	JARID2	ENSG00000008083	NON_SYNONYMOUS_CODING	ENST00000341776.2:c.3472G>A	ENSP00000341280.2:p.Val1158Ile	17/18	YES	-	-
31	SNV	chr6	15517413	G	A	527	287	240	0.455	JARID2	ENSG00000008083	NON_SYNONYMOUS_CODING	ENST00000397311.3:c.2956G>A	ENSP00000380478.3:p.Val986Ile	17/18	-	-	-
S12	SNV	chr6	15501627	A	G	291	144	147	0.505	JARID2	ENSG00000008083	NON_SYNONYMOUS_CODING	ENST00000341776.2:c.2435A>G	ENSP00000341280.2:p.Lys812Arg	8/18	YES	-	-
S12	SNV	chr6	15501627	A	G	291	144	147	0.505	JARID2	ENSG00000008083	NON_SYNONYMOUS_CODING	ENST00000397311.3:c.1919A>G	ENSP00000380478.3:p.Lys640Arg	8/18	-	-	-
S12	SNV	chr6	15501627	A	G	291	144	147	0.505	JARID2	ENSG00000008083	NON_SYNONYMOUS_CODING	ENST00000541660.1:c.2321A>G	ENSP00000444623.1:p.Lys774Arg	7/12	-	-	-
S17	INDEL	chr6	15497066	A	-G	718	204	514	0.716	JARID2	ENSG00000008083	FRAME_SHIFT	ENST00000541660.1:c.1497delG	ENSP00000444623.1:p.Asp500ThrfsTer52	6/12	-	-	-
S17	INDEL	chr6	15497066	A	-G	718	204	514	0.716	JARID2	ENSG00000008083	FRAME_SHIFT	ENST00000397311.3:c.1095delG	ENSP00000380478.3:p.Asp366ThrfsTer52	7/18	-	-	-
S17	INDEL	chr6	15497066	A	-G	718	204	514	0.716	JARID2	ENSG00000008083	FRAME_SHIFT	ENST00000341776.2:c.1611delG	ENSP00000341280.2:p.Asp538ThrfsTer52	7/18	YES	-	-
28	SNV	chr1	59248294	A	C	117	75	42	0.359	JUN	ENSG00000177606	NON_SYNONYMOUS_CODING	ENST00000371222.2:c.449T>G	ENSP00000360266.2:p.Val150Gly	1/1	YES	-	-
S40	INDEL	chrX	44966739	G	+C	554	321	233	0.421	KDM6A	ENSG00000147050	FRAME_SHIFT	-	-	16/18	-	-	-
S40	INDEL	chrX	44966739	G	+C	554	321	233	0.421	KDM6A	ENSG00000147050	FRAME_SHIFT	-	-	26/28	-	-	-
S40	INDEL	chrX	44966739	G	+C	554	321	233	0.421	KDM6A	ENSG00000147050	FRAME_SHIFT	-	-	15/17	-	-	-
S40	INDEL	chrX	44966739	G	+C	554	321	233	0.421	KDM6A	ENSG00000147050	FRAME_SHIFT	-	-	27/29	-	-	-
S40	INDEL	chrX	44966739	G	+C	554	321	233	0.421	KDM6A	ENSG00000147050	FRAME_SHIFT	-	-	27/29	YES	-	-
S40	INDEL	chrX	44966739	G	+C	554	321	233	0.421	KDM6A	ENSG00000147050	FRAME_SHIFT	-	-	25/27	-	-	-
S40	INDEL	chrX	44966739	G	+C	554	321	233	0.421	KDM6A	ENSG00000147050	FRAME_SHIFT	-	-	3/5	-	-	-
S06	SNV	chr4	55972900	A	G	440	232	208	0.473	KDR	ENSG00000128052	NON_SYNONYMOUS_CODING	ENST00000263923.4:c.1490T>C	ENSP00000263923.4:p.Ile497Thr	11/30	YES	-	COSM587587
1	SNV	chr14	105353184	A	G	332	185	147	0.443	KIAA0284	ENSG00000099814	NON_SYNONYMOUS_CODING	ENST00000418279.1:c.2398A>G	ENSP00000415006.1:p.Thr800Ala	11/18	-	-	-
1	SNV	chr14	105353184	A	G	332	185	147	0.443	KIAA0284	ENSG00000099814	NON_SYNONYMOUS_CODING	ENST00000453495.1:c.2611A>G	ENSP00000407238.1:p.Thr871Ala	12/19	-	-	-
1	SNV	chr14	105353184	A	G	332	185	147	0.443	KIAA0284	ENSG00000099814	NON_SYNONYMOUS_CODING	ENST00000414716.3:c.2608A>G	ENSP00000404151.2:p.Thr870Ala	12/19	YES	-	-
S47	SNV	chr14	105350774	C	A	157	79	78	0.497	KIAA0284	ENSG00000099814	NON_SYNONYMOUS_CODING	ENST00000453495.1:c.1661C>A	ENSP00000407238.1:p.Thr554Asn	9/19	-	-	-
S47	SNV	chr14	105350774	C	A	157	79	78	0.497	KIAA0284	ENSG00000099814	NON_SYNONYMOUS_CODING	ENST00000556508.1:c.1448C>A	ENSP00000451249.1:p.Thr483Asn	8/18	-	-	-
S47	SNV	chr14	105350774	C	A	157	79	78	0.497	KIAA0284	ENSG00000099814	NON_SYNONYMOUS_CODING	ENST00000414716.3:c.1658C>A	ENSP00000404151.2:p.Thr553Asn	9/19	YES	-	-
S47	SNV	chr14	105350774	C	A	157	79	78	0.497	KIAA0284	ENSG00000099814	NON_SYNONYMOUS_CODING	ENST00000418279.1:c.1448C>A	ENSP00000415006.1:p.Thr483Asn	8/18	-	-	-
S27	SNV	chr20	30898062	C	A	385	200	185	0.48	KIF3B	ENSG00000101350	NON_SYNONYMOUS_CODING	ENST00000375712.3:c.482C>A	ENSP00000364864.3:p.Thr161Asn	2/9	YES	-	-
S53	SNV	chr10	32322952	T	C	418	207	211	0.505	KIF5B	ENSG00000170759	NON_SYNONYMOUS_CODING	ENST00000302418.4:c.1126A>G	ENSP00000307078.4:p.Ile376Val	12/26	YES	0.0002	rs190199916
S40	SNV	chr4	55599340	T	G	613	315	298	0.486	KIT	ENSG00000157404	NON_SYNONYMOUS_CODING	ENST00000412167.2:c.2454T>G	ENSP00000390987.2:p.Asn818Lys	17/21	-	-	rs121913514,KinMutBase_KIT_DNA:g.76159T>A,COSM1321,COSM1320,COSM1322,COSM1319
S40	SNV	chr4	55599340	T	G	613	315	298	0.486	KIT	ENSG00000157404	NON_SYNONYMOUS_CODING	ENST00000288135.5:c.2466T>G	ENSP00000288135.5:p.Asn822Lys	17/21	YES	-	rs121913514,KinMutBase_KIT_DNA:g.76159T>A,COSM1321,COSM1320,COSM1322,COSM1319
24	SNV	chr13	33629468	A	T	298	148	150	0.503	KL	ENSG00000133116	STOP_GAINED	ENST00000426690.2:c.694A>T	ENSP00000399513.2:p.Lys232Ter	3/4	-	-	rs373674206
38	SNV	chr13	33590954	A	T	210	94	116	0.552	KL	ENSG00000133116	NON_SYNONYMOUS_CODING	ENST00000380099.3:c.376A>T	ENSP00000369442.3:p.Ser126Cys	1/5	YES	-	-
47	SNV	chr12	25380275	T	A	169	100	69	0.408	KRAS	ENSG00000133703	NON_SYNONYMOUS_CODING	ENST00000256078.4:c.183A>T	ENSP00000256078.4:p.Gln61His	3/6	YES	-	rs17851045,COSM555,COSM554,COSM1135364,COSM1146992
47	SNV	chr12	25380275	T	A	169	100	69	0.408	KRAS	ENSG00000133703	NON_SYNONYMOUS_CODING	ENST00000311936.3:c.183A>T	ENSP00000308495.3:p.Gln61His	3/5	-	-	rs17851045,COSM555,COSM554,COSM1135364,COSM1146992
S23	SNV	chr12	25398284	C	T	513	283	230	0.448	KRAS	ENSG00000133703	NON_SYNONYMOUS_CODING	ENST00000556131.1:c.35G>A	ENSP00000451856.1:p.Gly12Asp	2/3	-	-	COSM520,COSM521,COSM522,COSM12657,COSM1140133,COSM1135366,COSM1140134,CM087372
S23	SNV	chr12	25398284	C	T	513	283	230	0.448	KRAS	ENSG00000133703	NON_SYNONYMOUS_CODING	ENST00000311936.3:c.35G>A	ENSP00000308495.3:p.Gly12Asp	2/5	-	-	COSM520,COSM521,COSM522,COSM12657,COSM1140133,COSM1135366,COSM1140134,CM087372

S23	SNV	chr12	25398284	C	T	513	283	230	0.448	KRAS	ENSG00000133703	NON_SYNONYMOUS_CODING	ENST00000256078.4:c.35G>A	ENSP00000256078.4:p.Gly12Asp	2/6	YES	-	COSM520,COSM521,COSM522,COSM12657,COSM1140133,COSM1135366,COSM1140134,CM087372
S23	SNV	chr12	25398284	C	T	513	283	230	0.448	KRAS	ENSG00000133703	NON_SYNONYMOUS_CODING	ENST00000557334.1:c.35G>A	ENSP00000452512.1:p.Gly12Asp	2/3	-	-	COSM520,COSM521,COSM522,COSM12657,COSM1140133,COSM1135366,COSM1140134,CM087372
58	SNV	chr9	133944333	G	A	183	100	83	0.454	LAMC3	ENSG00000050555	NON_SYNONYMOUS_CODING	ENST00000361069.4:c.2786G>A	ENSP00000354360.4:p.Cys929Tyr	16/28	YES	-	-
S30	SNV	chr1	156104615	G	A	421	331	90	0.214	LMNA	ENSG00000160789	NON_SYNONYMOUS_CODING	ENST00000361308.4:c.659G>A	ENSP00000355292.4:p.Arg220His	4/10	-	-	-
S30	SNV	chr1	156104615	G	A	421	331	90	0.214	LMNA	ENSG00000160789	NON_SYNONYMOUS_CODING	ENST00000473598.2:c.362G>A	ENSP00000421821.1:p.Arg121His	5/13	-	-	-
S30	SNV	chr1	156104615	G	A	421	331	90	0.214	LMNA	ENSG00000160789	NON_SYNONYMOUS_CODING	ENST00000368299.3:c.659G>A	ENSP00000357282.3:p.Arg220His	4/12	-	-	-
S30	SNV	chr1	156104615	G	A	421	331	90	0.214	LMNA	ENSG00000160789	NON_SYNONYMOUS_CODING	ENST00000392353.3:c.416G>A	ENSP00000376164.3:p.Arg139His	4/10	-	-	-
S30	SNV	chr1	156104615	G	A	421	331	90	0.214	LMNA	ENSG00000160789	NON_SYNONYMOUS_CODING	ENST00000368297.1:c.416G>A	ENSP00000357280.1:p.Arg139His	5/11	-	-	-
S30	SNV	chr1	156104615	G	A	421	331	90	0.214	LMNA	ENSG00000160789	NON_SYNONYMOUS_CODING	ENST00000448611.2:c.323G>A	ENSP00000395597.2:p.Arg108His	4/13	-	-	-
S30	SNV	chr1	156104615	G	A	421	331	90	0.214	LMNA	ENSG00000160789	NON_SYNONYMOUS_CODING	ENST00000368300.4:c.659G>A	ENSP00000357283.4:p.Arg220His	4/12	YES	-	-
S30	SNV	chr1	156104615	G	A	421	331	90	0.214	LMNA	ENSG00000160789	NON_SYNONYMOUS_CODING	ENST00000347559.2:c.659G>A	ENSP00000292304.3:p.Arg220His	4/11	-	-	-
S30	SNV	chr1	156104615	G	A	421	331	90	0.214	LMNA	ENSG00000160789	NON_SYNONYMOUS_CODING	ENST00000368301.2:c.659G>A	ENSP00000357284.2:p.Arg220His	7/13	-	-	-
S30	SNV	chr1	156104615	G	A	421	331	90	0.214	LMNA	ENSG00000160789	NON_SYNONYMOUS_CODING	ENST00000504687.1:c.410G>A	ENSP00000426535.1:p.Arg137His	4/4	-	-	-
S43	SNV	chr11	33886286	C	T	438	235	203	0.464	LMO2	ENSG00000135363	NON_SYNONYMOUS_CODING	ENST00000395833.3:c.119G>A	ENSP00000379175.3:p.Arg40His	2/3	-	-	-
S43	SNV	chr11	33886286	C	T	438	235	203	0.464	LMO2	ENSG00000135363	NON_SYNONYMOUS_CODING	ENST00000257818.2:c.328G>A	ENSP00000257818.2:p.Arg110His	5/6	YES	-	-
8	SNV	chr3	188327502	G	A	1252	471	781	0.624	LPP	ENSG00000145012	NON_SYNONYMOUS_CODING	ENST00000448637.1:c.983G>A	ENSP00000393602.1:p.Arg328Gln	6/7	-	-	-
8	SNV	chr3	188327502	G	A	1252	471	781	0.624	LPP	ENSG00000145012	NON_SYNONYMOUS_CODING	ENST00000543006.1:c.983G>A	ENSP00000438891.1:p.Arg328Gln	6/11	-	-	-
8	SNV	chr3	188327502	G	A	1252	471	781	0.624	LPP	ENSG00000145012	NON_SYNONYMOUS_CODING	ENST00000312675.4:c.983G>A	ENSP00000318089.4:p.Arg328Gln	6/11	YES	-	-
8	SNV	chr3	188327502	G	A	1252	471	781	0.624	LPP	ENSG00000145012	NON_SYNONYMOUS_CODING	ENST00000415906.1:c.494G>A	ENSP00000393008.1:p.Arg165Gln	1/3	-	-	-
6	SNV	chr12	59271268	C	G	613	325	288	0.47	LRIG3	ENSG00000139263	NON_SYNONYMOUS_CODING	ENST00000320743.3:c.2450G>C	ENSP00000326759.3:p.Cys817Ser	15/19	YES	-	COSM1285970
6	SNV	chr12	59271268	C	G	613	325	288	0.47	LRIG3	ENSG00000139263	NON_SYNONYMOUS_CODING	ENST00000379141.4:c.2270G>C	ENSP00000368436.4:p.Cys757Ser	15/19	-	-	COSM1285970
S10	SNV	chr12	59274576	T	C	771	381	390	0.506	LRIG3	ENSG00000139263	NON_SYNONYMOUS_CODING	ENST00000320743.3:c.1588A>G	ENSP00000326759.3:p.Met530Val	13/19	YES	-	-
S10	SNV	chr12	59274576	T	C	771	381	390	0.506	LRIG3	ENSG00000139263	NON_SYNONYMOUS_CODING	ENST00000379141.4:c.1408A>G	ENSP00000368436.4:p.Met470Val	13/19	-	-	-
S16	SNV	chr12	59314010	C	A	69	40	29	0.42	LRIG3	ENSG00000139263	NON_SYNONYMOUS_CODING	ENST00000320743.3:c.7G>T	ENSP00000326759.3:p.Ala3Ser	1/19	YES	-	-
S65	SNV	chr12	59274476	G	T	744	357	387	0.52	LRIG3	ENSG00000139263	NON_SYNONYMOUS_CODING	ENST00000379141.4:c.1508C>A	ENSP00000368436.4:p.Thr503Asn	13/19	-	-	-
S65	SNV	chr12	59274476	G	T	744	357	387	0.52	LRIG3	ENSG00000139263	NON_SYNONYMOUS_CODING	ENST00000320743.3:c.1688C>A	ENSP00000326759.3:p.Thr563Asn	13/19	YES	-	-
19	SNV	chr17	11984813	T	C	510	270	240	0.471	MAP2K4	ENSG00000065559	NON_SYNONYMOUS_CODING	ENST00000353533.5:c.359T>C	ENSP00000262445.5:p.Val120Ala	3/11	YES	-	-
19	SNV	chr17	11984813	T	C	510	270	240	0.471	MAP2K4	ENSG00000065559	NON_SYNONYMOUS_CODING	ENST00000415385.3:c.392T>C	ENSP00000410402.3:p.Val131Ala	4/12	-	-	-
25	SNV	chr1	23956027	C	T	278	154	124	0.446	MED12	ENSG00000197880	UTR_5_PRIME	ENST00000374555.3:c.-18C>T	-	4/7	YES	-	rs372767616
S19	SNV	chrX	70355014	C	T	275	41	234	0.851	MED12	ENSG00000184634	STOP_GAINED	ENST00000374102.1:c.4936C>T	ENSP00000363215.1:p.Arg1646Ter	36/45	-	-	COSM3845293,COSM3845294
S19	SNV	chrX	70355014	C	T	275	41	234	0.851	MED12	ENSG00000184634	STOP_GAINED	ENST00000333646.6:c.4936C>T	ENSP00000333125.6:p.Arg1646Ter	36/45	-	-	COSM3845293,COSM3845294
S19	SNV	chrX	70355014	C	T	275	41	234	0.851	MED12	ENSG00000184634	STOP_GAINED	ENST00000374080.3:c.4936C>T	ENSP00000363193.3:p.Arg1646Ter	36/45	YES	-	COSM3845293,COSM3845294
47	SNV	chr7	116340189	C	G	249	106	143	0.574	MET	ENSG00000105976	NON_SYNONYMOUS_CODING	ENST00000397752.3:c.1051C>G	ENSP00000380860.3:p.Pro351Ala	2/21	-	-	-
47	SNV	chr7	116340189	C	G	249	106	143	0.574	MET	ENSG00000105976	NON_SYNONYMOUS_CODING	ENST00000436117.2:c.1051C>G	ENSP00000410982.2:p.Pro351Ala	1/9	-	-	-
47	SNV	chr7	116340189	C	G	249	106	143	0.574	MET	ENSG00000105976	NON_SYNONYMOUS_CODING	ENST00000318493.6:c.1051C>G	ENSP00000317272.6:p.Pro351Ala	2/21	YES	-	-
S10	INDEL	chr22	40815141	G	-CCA	85	52	33	0.388	MKL1	ENSG00000196588	CODON_CHANGE_PLUS_CODON_DELETION	ENST00000355630.3:c.1302delC	ENSP00000347847.3:p.Thr435Arg fsTer8	12/15	YES	0.0002	rs149045760
S10	INDEL	chr22	40815141	G	-CCA	85	52	33	0.388	MKL1	ENSG00000196588	CODON_CHANGE_PLUS_CODON_DELETION	ENST00000396617.3:c.1302delC	ENSP00000379861.3:p.Thr435Arg fsTer8	12/15	-	0.0002	rs149045760
S10	INDEL	chr22	40815141	G	-CCA	85	52	33	0.388	MKL1	ENSG00000196588	CODON_CHANGE_PLUS_CODON_DELETION	ENST00000402042.1:c.1152delC	ENSP00000385584.1:p.Thr385Arg fsTer8	11/14	-	0.0002	rs149045760
S10	INDEL	chr22	40815141	G	-CCA	85	52	33	0.388	MKL1	ENSG00000196588	CODON_CHANGE_PLUS_CODON_DELETION	ENST00000407029.1:c.1302delC	ENSP00000385835.1:p.Thr435Arg fsTer8	9/12	-	0.0002	rs149045760
37	SNV	chr3	37089011	A	G	32	20	12	0.375	MLH1	ENSG00000076242	NON_SYNONYMOUS_CODING	ENST00000539477.1:c.1010A>G	ENSP00000443665.1:p.Glu337Gly	15/18	-	-	rs63751612,MMR_c.1733A>G,CM950806
37	SNV	chr3	37089011	A	G	32	20	12	0.375	MLH1	ENSG00000076242	NON_SYNONYMOUS_CODING	ENST00000435176.1:c.1439A>G	ENSP00000402564.1:p.Glu480Gly	16/19	-	-	rs63751612,MMR_c.1733A>G,CM950806
37	SNV	chr3	37089011	A	G	32	20	12	0.375	MLH1	ENSG00000076242	NON_SYNONYMOUS_CODING	ENST00000536378.1:c.1010A>G	ENSP00000444286.1:p.Glu337Gly	16/19	-	-	rs63751612,MMR_c.1733A>G,CM950806
37	SNV	chr3	37089011	A	G	32	20	12	0.375	MLH1	ENSG00000076242	NON_SYNONYMOUS_CODING	ENST00000455445.2:c.1010A>G	ENSP00000398272.2:p.Glu337Gly	16/19	-	-	rs63751612,MMR_c.1733A>G,CM950806
37	SNV	chr3	37089011	A	G	32	20	12	0.375	MLH1	ENSG00000076242	NON_SYNONYMOUS_CODING	ENST00000458205.2:c.1010A>G	ENSP00000402967.2:p.Glu337Gly	17/20	-	-	rs63751612,MMR_c.1733A>G,CM950806
37	SNV	chr3	37089011	A	G	32	20	12	0.375	MLH1	ENSG00000076242	NON_SYNONYMOUS_CODING	ENST00000231790.2:c.1733A>G	ENSP00000231790.2:p.Glu578Gly	16/19	YES	-	rs63751612,MMR_c.1733A>G,CM950806
47	SNV	chr11	118344095	T	A	310	152	158	0.51	MLL	ENSG00000118058	NON_SYNONYMOUS_CODING	ENST00000531904.2:c.2320T>A	ENSP00000432391.2:p.Phe774Ile	4/10	-	-	-
47	SNV	chr11	118344095	T	A	310	152	158	0.51	MLL	ENSG00000118058	NON_SYNONYMOUS_CODING	ENST00000534358.1:c.2221T>A	ENSP00000436786.1:p.Phe741Ile	3/36	YES	-	-
47	SNV	chr11	118344095	T	A	310	152	158	0.51	MLL	ENSG00000118058	NON_SYNONYMOUS_CODING	ENST00000354520.4:c.2221T>A	ENSP00000346516.4:p.Phe741Ile	3/35	-	-	-
47	SNV	chr11	118344095	T	A	310	152	158	0.51	MLL	ENSG00000118058	NON_SYNONYMOUS_CODING	ENST00000389506.5:c.2221T>A	ENSP00000374157.5:p.Phe741Ile	3/36	-	-	-
S59	SNV	chr11	118373257	G	A	696	339	357	0.513	MLL	ENSG00000118058	NON_SYNONYMOUS_CODING	ENST00000534358.1:c.6650G>A	ENSP00000436786.1:p.Arg2217Lys	27/36	YES	-	-
S59	SNV	chr11	118373257	G	A	696	339	357	0.513	MLL	ENSG00000118058	NON_SYNONYMOUS_CODING	ENST00000389506.5:c.6641G>A	ENSP00000374157.5:p.Arg2214Lys	27/36	-	-	-
S59	SNV	chr11	118373257	G	A	696	339	357	0.513	MLL	ENSG00000118058	NON_SYNONYMOUS_CODING	ENST00000354520.4:c.6527G>A	ENSP00000346516.4:p.Arg2176Lys	26/35	-	-	-
11	SNV	chr12	49427360	C	T	179	111	68	0.38	MLL2	ENSG00000167548	NON_SYNONYMOUS_CODING	ENST00000301067.7:c.11128G>A	ENSP00000301067.7:p.Gly3710Arg	39/54	YES	-	-
12	INDEL	chr12	49427265	T	+TGC	170	113	57	0.331	MLL2	ENSG00000167548	CODON_CHANGE_PLUS_CODON_INSERTION	-	-	39/54	YES	-	-
35	INDEL	chr12	49427265	T	+TGC	217	142	75	0.346	MLL2	ENSG00000167548	CODON_CHANGE_PLUS_CODON_INSERTION	-	-	39/54	YES	-	-
7	SNV	chr12	49445391	G	C	324	177	147	0.454	MLL2	ENSG00000167548	NON_SYNONYMOUS_CODING	ENST00000301067.7:c.2075C>G	ENSP00000301067.7:p.Pro692Arg	10/54	YES	-	-
S45	SNV	chr12	49447286	G	C	403	209	194	0.481	MLL2	ENSG00000167548	NON_SYNONYMOUS_CODING	ENST00000301067.7:c.812C>G	ENSP00000301067.7:p.Pro271Arg	6/54	YES	-	-
1	SNV	chr7	151877921	T	C	281	157	124	0.441	MLL3	ENSG00000055609	NON_SYNONYMOUS_CODING	ENST00000262189.6:c.7024A>G	ENSP00000262189.6:p.Met2342Val	36/59	YES	-	-
1	SNV	chr7	151877921	T	C	281	157	124	0.441	MLL3	ENSG00000055609	NON_SYNONYMOUS_CODING	ENST00000355193.2:c.7024A>G	ENSP00000347325.2:p.Met2342Val	36/60	-	-	-
S15	SNV	chr7	151932991	G	A	453	350	103	0.227	MLL3	ENSG00000055609	NON_SYNONYMOUS_CODING	ENST00000262189.6:c.2680C>T	ENSP00000262189.6:p.Arg894Trp	16/59	YES	-	-
S15	SNV	chr7	151932991	G	A	453	350	103	0.227	MLL3	ENSG00000055609	NON_SYNONYMOUS_CODING	ENST00000418673.1:c.146C>T	ENSP00000403483.1:p.Arg50Trp	2/6	-	-	-
S15	SNV	chr7	151932991	G	A	453	350	103	0.227	MLL3	ENSG00000055609	NON_SYNONYMOUS_CODING	ENST00000355193.2:c.2680C>T	ENSP00000347325.2:p.Arg894Trp	16/60	-	-	-
S22	INDEL	chr9	20414400	G	+CTG	85	61	24	0.279	MLLT3	ENSG00000171843	CODON_INSERTION	-	-	5/11	YES	-	-
S22	INDEL	chr9	20414400	G	+CTG	85	61	24	0.279	MLLT3	ENSG00000171843	CODON_INSERTION	-	-	5/11	-	-	-
S43	SNV	chr9	20448225	G	A	366	192	174	0.475	MLLT3	ENSG00000171843	START_GAINED	ENST00000355930.6:c.-903C>T	-	4/10	-	-	COSM3942994
S05	SNV	chr6	168347561	G	A	350	192	158	0.451	MLLT4	ENSG00000130396	NON_SYNONYMOUS_CODING	ENST00000400822.3:c.3509G>A	ENSP00000383623.3:p.Arg1170His	26/34	-	-	-
S05	SNV	chr6	168347561	G	A	350	192	158	0.451	MLLT4	ENSG00000130396	NON_SYNONYMOUS_CODING	ENST00000392112.1:c.3461G>A	ENSP00000375960.1:p.Arg1154His	26/33	-	-	-
S05	SNV	chr6	168347561	G	A	350	192	158	0.451	MLLT4	ENSG00000130396	NON_SYNONYMOUS_CODING	ENST00000351017.4:c.3533G>A	ENSP00000252692.5:p.Arg1178His	27/34	-	-	-
S05	SNV	chr6	168347561	G	A	350	192	158	0.451	MLLT4	ENSG00000130396	NON_SYNONYMOUS_CODING	ENST00000447894.2:c.3512G>A	ENSP00000404595.2:p.Arg1171His	26/33	-	-	-
S05	SNV	chr6	168347561	G	A	350	192	158	0.451	MLLT4	ENSG00000130396	NON_SYNONYMOUS_CODING	ENST00000366806.2:c.3512G>A	ENSP00000355771.2:p.Arg1171His	27/34	-	-	-
S05	SNV	chr6	168347561	G	A	350	192	158	0.451	MLLT4	ENSG00000130396	NON_SYNONYMOUS_CODING	ENST00000392108.3:c.3512G>A	ENSP00000375956.3:p.Arg1171His	26/30	YES	-	-
S05	SNV	chr6	168347561	G	A	350	192	158	0.451	MLLT4	ENSG00000130396	NON_SYNONYMOUS_CODING	ENST00000344191.4:c.3512G>A	ENSP00000341118.4:p.Arg1171His	26/31	-	-	-
S34	INDEL	chr6	168366912	C	+A	373	205	168	0.45	MLLT4	ENSG00000130396	FRAME_SHIFT	-	-	1/3	-	-	-
S34	INDEL	chr6	168366912	C	+A	373	205	168	0.45	MLLT4	ENSG00000130396	FRAME_SHIFT	-	-	1/3	-	-	-
S38	SNV	chr6	168366557	C	T	114	53	61	0.535	MLLT4	ENSG00000130396	NON_SYNONYMOUS_CODING	ENST00000447894.2:c.5068C>T	ENSP00000404595.2:p.Arg1690Trp	31/33	-	-	COSM1173200
S38	SNV	chr6	168366557	C	T	114	53	61	0.535	MLLT4	ENSG00000130396	NON_SYNONYMOUS_CODING	ENST00000351017.4:c.5089C>T	ENSP00000252692.5:p.Arg1697Trp	32/34	-	-	COSM1173200
S38	SNV	chr6	168366557	C	T	114	53	61	0.535	MLLT4	ENSG00000130396	NON_SYNONYMOUS_CODING	ENST00000507704.1:c.534C>T	ENSP00000421441.1:p.Arg179Trp	3/5	-	-	COSM1173200
S38	SNV	chr6	168366557	C	T	114	53	61	0.535	MLLT4	ENSG00000130396	NON_SYNONYMOUS_CODING	ENST00000400822.3:c.5098C>T	ENSP00000383623.3:p.Arg1700Trp	32/34	-	-	COSM1173200
S38	SNV	chr6	168366557	C	T	114	53	61	0.535	MLLT4	ENSG00000130396	NON_SYNONYMOUS_CODING	ENST00000366806.2:c.5068C>T	ENSP00000355771.2:p.Arg1690Trp	32/34	-	-	COSM1173200
S42	SNV	chr6	168352525	C	G	584	303	281	0.481	MLLT4	ENSG00000130396	NON_SYNONYMOUS_CODING	ENST00000392112.1:c.4419C>G	ENSP00000375960.1:p.Ile1473Met	29/33	-	-	rs377069127
S42	SNV	chr6	168352525	C	G	584	303	281	0.481	MLLT4	ENSG00000130396	NON_SYNONYMOUS_CODING	ENST00000366806.2:c.4470C>G	ENSP00000355771.2:p.Ile1490Met	30/34	-	-	rs377069127
S42	SNV	chr6	168352525	C	G	584	303	281	0.481	MLLT4	ENSG00000130396	NON_SYNONYMOUS_CODING	ENST00000400822.3:c.4467C>G	ENSP00000383623.3:p.Ile1489Met	29/34	-	-	rs377069127
S42	SNV	chr6	168352525	C	G	584	303	281	0.481	MLLT4	ENSG00000130396	NON_SYNONYMOUS_CODING	ENST00000351017.4:c.4491C>G	ENSP00000252692.5:p.Ile1497Met	30/34	-	-	rs377069127
S42	SNV	chr6	168352525	C	G	584	303	281	0.481	MLLT4	ENSG00000130396	NON_SYNONYMOUS_CODING	ENST00000447894.2:c.4470C>G	ENSP00000404595.2:p.Ile1490Met	29/33	-	-	rs377069127
S42	SNV	chr6	168352525	C	G	584	303	281	0.481	MLLT4	ENSG00000130396	NON_SYNONYMOUS_CODING	ENST00000344191.4:c.4470C>G	ENSP00000341118.4:p.Ile1490Met	29/31	-	-	rs377069127
S42	SNV	chr6	168352525	C	G	584	303	281	0.481	MLLT4	ENSG00000130396	NON_SYNONYMOUS_CODING	ENST00000392108.3:c.4470C>G	ENSP00000375956.3:p.Ile1490Met	29/30	YES	-	rs377069127
S48	SNV	chr6	168276029	T	C	546	398	148	0.271	MLLT4	ENSG00000130396	NON_SYNONYMOUS_CODING	ENST00000400825.4:c.593T>C	ENSP00000383625.4:p.Leu198Pro	5/6	-	-	-
S48	SNV	chr6	168276029	T	C	546	398	148	0.271	MLLT4	ENSG00000130396	NON_SYNONYMOUS_CODING	ENST00000392108.3:c.593T>C	ENSP00000375956.3:p.Leu198Pro	5/30	YES	-	-
S48	SNV	chr6	168276029	T	C	546	398	148	0.271	MLLT4	ENSG00000130396	NON_SYNONYMOUS_CODING	ENST00000344191.4:c.593T>C	ENSP00000341118.4:p.Leu198Pro	5/31	-	-	-
S48	SNV	chr6	168276029	T	C	546	398	148	0.271	MLLT4	ENSG00000130396	NON_SYNONYMOUS_CODING	ENST00000447894.2:c.593T>C	ENSP00000404595.2:p.Leu198Pro	5/33	-	-	-
S48	SNV	chr6	168276029	T	C	546	398	148	0.271	MLLT4	ENSG00000130396	NON_SYNONYMOUS_CODING	ENST00000392112.1:c.590T>C	ENSP00000375960.1:p.Leu197Pro	5/33	-	-	-
S48	SNV	chr6	168276029	T	C	546	398	148	0.271	MLLT4	ENSG00000130396	NON_SYNONYMOUS_CODING	ENST00000366806.2:c.593T>C	ENSP00000355771.2:p.Leu198Pro	5/34	-	-	-
S48	SNV	chr6	168276029	T	C	546	398	148	0.271	MLLT4	ENSG00000130396	NON_SYNONYMOUS_CODING	ENST00000400822.3:c.590T>C	ENSP00000383623.3:p.Leu197Pro	5/34	-	-	-
S48	SNV	chr6	168276029	T	C	546	398	148	0.271	MLLT4	ENSG00000130396	NON_SYNONYMOUS_CODING	ENST00000351017.4:c.593T>C	ENSP00000252692.5:p.Leu198Pro	5/34	-	-	-
S48	SNV	chr6	168276029	T	C	546	398	148	0.271	MLLT4	ENSG00000130396	NON_SYNONYMOUS_CODING	ENST00000400824.4:c.593T>C	ENSP00000383625.4:p.Leu199Pro	5/5	-	-	-
S52	SNV	chr6	168366779	C	T	521	255	266	0.511	MLLT4	ENSG00000130396	NON_SYNONYMOUS_CODING	ENST00000485634.2:c.164C>T	ENSP00000423035.1:p.Pro56Ser	1/3	-	-	-
25	SNV	chr22	28193237	G	C	74	43	31	0.419	MN1	ENSG00000169184	NON_SYNONYMOUS_CODING	ENST00000302326.4:c.3295C>G	ENSP00000304956.4:p.Pro1099Ala	1/2	YES	0.0002	rs560561226

25	SNV	chr22	28193598	C	A	156	87	69	0.442	MN1	ENSG00000169184	NON_SYNONYMOUS_CODING	ENST00000302326.4:c.2934G>T	ENSP00000304956.4:p.Gln978His	1/2	YES	0.0010	rs140987101	
S05	INDEL	chr22	28194188	C	GCCACCGCCGCCAC CAGAGCT	38	25	13	0.342	MN1	ENSG00000169184	CODON_CHANGE_PLUS_CODON_DELETION	ENST00000302326.4:c.2348delG	ENSP00000304956.4:p.Gly783ValfsTer53	1/2	YES	-	-	
21	SNV	chr7	156798348	C	T	111	67	44	0.396	MNX1	ENSG00000130675	NON_SYNONYMOUS_CODING	ENST00000543409.1:c.436G>A	ENSP00000438552.1:p.Glu146Lys	3/3	-	-	-	
21	SNV	chr7	156798348	C	T	111	67	44	0.396	MNX1	ENSG00000130675	NON_SYNONYMOUS_CODING	ENST00000252971.6:c.1072G>A	ENSP00000252971.5:p.Glu358Lys	3/3	YES	-	-	
S22	SNV	chr7	156802466	G	C	98	49	49	0.5	MNX1	ENSG00000130675	NON_SYNONYMOUS_CODING	ENST00000252971.6:c.579C>G	ENSP00000252971.5:p.His193Gln	1/3	YES	-	-	
S20	SNV	chr1	43818309	C	T	1070	626	444	0.415	MPL	ENSG00000117400	STOP_GAINED	ENST00000372470.3:c.1774C>T	ENSP00000361548.3:p.Arg592Ter	12/12	YES	-	COSM366895	
S20	SNV	chr1	43804323	A	T	1058	637	421	0.398	MPL	ENSG00000117400	NON_SYNONYMOUS_CODING	ENST00000413094.2:c.323A>T	ENSP00000414004.2:p.His108Leu	3/10	-	-	-	
S20	SNV	chr1	43804323	A	T	1058	637	421	0.398	MPL	ENSG00000117400	NON_SYNONYMOUS_CODING	ENST00000372470.3:c.323A>T	ENSP00000361548.3:p.His108Leu	3/12	YES	-	-	
38	SNV	chr2	48027478	T	A	766	373	393	0.513	MSH6	ENSG00000116062	NON_SYNONYMOUS_CODING	ENST00000540021.1:c.1966T>A	ENSP00000446475.1:p.Tyr656Asn	2/8	-	-	-	
38	SNV	chr2	48027478	T	A	766	373	393	0.513	MSH6	ENSG00000116062	NON_SYNONYMOUS_CODING	ENST00000234420.5:c.2356T>A	ENSP00000234420.4:p.Tyr786Asn	4/10	YES	-	-	
38	SNV	chr2	48027478	T	A	766	373	393	0.513	MSH6	ENSG00000116062	NON_SYNONYMOUS_CODING	ENST00000538136.1:c.1450T>A	ENSP00000438580.1:p.Tyr484Asn	4/10	-	-	-	
21	SNV	chr1	11190824	G	A	245	143	102	0.416	MTOR	ENSG00000198793	UTR_5_PRIME	ENST00000376838.1:c.-11C>T		1/20	-	-	COSM1215724	
21	SNV	chr1	11190824	G	A	245	143	102	0.416	MTOR	ENSG00000198793	NON_SYNONYMOUS_CODING	ENST00000361445.4:c.5375C>T	ENSP00000354558.4:p.Ala1792Val	39/58	YES	-	COSM1215724	
6	SNV	chr1	11166761	T	C	793	411	382	0.482	MTOR	ENSG00000198793	UTR_3_PRIME	ENST00000361445.4:c.*781A>G		58/58	YES	-	-	
6	SNV	chr1	11166761	T	C	793	411	382	0.482	MTOR	ENSG00000198793	UTR_3_PRIME	ENST00000376838.1:c.*781A>G	-	20/20	-	-	-	
S12	SNV	chr1	11272403	G	A	453	253	200	0.442	MTOR	ENSG00000198793	NON_SYNONYMOUS_CODING	ENST00000361445.4:c.3527C>T	ENSP00000354558.4:p.Thr1176Met	23/58	YES	0.0002	rs557533784	
35	INDEL	chr6	135515508	T	-G	319	197	122	0.381	MYB	ENSG00000118513	FRAME_SHIFT	ENST00000367814.4:c.859delG	ENSP00000356788.4:p.Glu287Lys	8/15	-	-	-	
35	INDEL	chr6	135515508	T	-G	319	197	122	0.381	MYB	ENSG00000118513	FRAME_SHIFT	ENST00000442647.2:c.859delG	ENSP00000410825.2:p.Glu287Lys	8/15	-	-	-	
35	INDEL	chr6	135515508	T	-G	319	197	122	0.381	MYB	ENSG00000118513	FRAME_SHIFT	ENST00000527615.1:c.859delG	ENSP00000433227.1:p.Glu287Lys	8/14	-	-	-	
35	INDEL	chr6	135515508	T	-G	319	197	122	0.381	MYB	ENSG00000118513	FRAME_SHIFT	ENST00000430686.2:c.721delG	ENSP00000390460.2:p.Glu241Lys	7/7	-	-	-	
35	INDEL	chr6	135515508	T	-G	319	197	122	0.381	MYB	ENSG00000118513	FRAME_SHIFT	ENST00000420123.2:c.787delG	ENSP00000403387.2:p.Glu263Lys	7/7	-	-	-	
35	INDEL	chr6	135515508	T	-G	319	197	122	0.381	MYB	ENSG00000118513	FRAME_SHIFT	ENST00000341911.5:c.859delG	ENSP00000339992.5:p.Glu287Lys	8/16	YES	-	-	
35	INDEL	chr6	135515508	T	-G	319	197	122	0.381	MYB	ENSG00000118513	FRAME_SHIFT	ENST00000534121.1:c.859delG	ENSP00000432851.1:p.Glu287Lys	8/16	-	-	-	
35	INDEL	chr6	135515508	T	-G	319	197	122	0.381	MYB	ENSG00000118513	FRAME_SHIFT	ENST00000534044.1:c.859delG	ENSP00000435055.1:p.Glu287Lys	8/14	-	-	-	
35	INDEL	chr6	135515508	T	-G	319	197	122	0.381	MYB	ENSG00000118513	FRAME_SHIFT	ENST00000316528.8:c.859delG	ENSP00000326328.8:p.Glu287Lys	8/17	-	-	-	
35	INDEL	chr6	135515508	T	-G	319	197	122	0.381	MYB	ENSG00000118513	FRAME_SHIFT	ENST00000525369.1:c.859delG	ENSP00000435938.1:p.Glu287Lys	8/14	-	-	-	
35	INDEL	chr6	135515508	T	-G	319	197	122	0.381	MYB	ENSG00000118513	FRAME_SHIFT	ENST00000528774.1:c.859delG	ENSP00000434723.1:p.Glu287Lys	8/16	-	-	-	
47	INDEL	chr6	135513580	A	-AG	301	178	123	0.409	MYB	ENSG00000118513	FRAME_SHIFT	ENST00000420123.2:c.575delA	ENSP00000403387.2:p.Lys192ArgfsTer5	5/7	-	-	-	
47	INDEL	chr6	135513580	A	-AG	301	178	123	0.409	MYB	ENSG00000118513	FRAME_SHIFT	ENST00000367814.4:c.647delA	ENSP00000356788.4:p.Lys216ArgfsTer5	6/15	-	-	-	
47	INDEL	chr6	135513580	A	-AG	301	178	123	0.409	MYB	ENSG00000118513	FRAME_SHIFT	ENST00000430686.2:c.509delA	ENSP00000390460.2:p.Lys170ArgfsTer5	5/7	-	-	-	
47	INDEL	chr6	135513580	A	-AG	301	178	123	0.409	MYB	ENSG00000118513	FRAME_SHIFT	ENST00000316528.8:c.647delA	ENSP00000326328.8:p.Lys216ArgfsTer5	6/17	-	-	-	
47	INDEL	chr6	135513580	A	-AG	301	178	123	0.409	MYB	ENSG00000118513	FRAME_SHIFT	ENST00000341911.5:c.647delA	ENSP00000339992.5:p.Lys216ArgfsTer5	6/16	YES	-	-	
47	INDEL	chr6	135513580	A	-AG	301	178	123	0.409	MYB	ENSG00000118513	FRAME_SHIFT	ENST00000442647.2:c.647delA	ENSP00000410825.2:p.Lys216ArgfsTer5	6/15	-	-	-	
47	INDEL	chr6	135513580	A	-AG	301	178	123	0.409	MYB	ENSG00000118513	FRAME_SHIFT	ENST00000527615.1:c.647delA	ENSP00000433227.1:p.Lys216ArgfsTer5	6/14	-	-	-	
47	INDEL	chr6	135513580	A	-AG	301	178	123	0.409	MYB	ENSG00000118513	FRAME_SHIFT	ENST00000525369.1:c.647delA	ENSP00000435938.1:p.Lys216ArgfsTer5	6/14	-	-	-	
47	INDEL	chr6	135513580	A	-AG	301	178	123	0.409	MYB	ENSG00000118513	FRAME_SHIFT	ENST00000534044.1:c.647delA	ENSP00000435055.1:p.Lys216ArgfsTer5	6/14	-	-	-	
47	INDEL	chr6	135513580	A	-AG	301	178	123	0.409	MYB	ENSG00000118513	FRAME_SHIFT	ENST00000533624.1:c.647delA	ENSP00000436605.1:p.Lys216ArgfsTer5	6/14	-	-	-	
47	INDEL	chr6	135513580	A	-AG	301	178	123	0.409	MYB	ENSG00000118513	FRAME_SHIFT	ENST00000528774.1:c.647delA	ENSP00000434723.1:p.Lys216ArgfsTer5	6/16	-	-	-	
47	INDEL	chr6	135513580	A	-AG	301	178	123	0.409	MYB	ENSG00000118513	FRAME_SHIFT	ENST00000534121.1:c.647delA	ENSP00000432851.1:p.Lys216ArgfsTer5	6/16	-	-	-	
S54	SNV	chr1	40366674	G	C	202	99	103	0.51	MYCL	ENSG00000116990	NON_SYNONYMOUS_CODING	ENST00000429311.1:c.433C>G	ENSP00000389358.1:p.Pro145Ala	2/2	-	-	-	
S54	SNV	chr1	40366674	G	C	202	99	103	0.51	MYCL	ENSG00000116990	NON_SYNONYMOUS_CODING	ENST00000372816.2:c.433C>G	ENSP00000361903.2:p.Pro145Ala	1/2	-	-	-	
S54	SNV	chr1	40366674	G	C	202	99	103	0.51	MYCL	ENSG00000116990	NON_SYNONYMOUS_CODING	ENST00000397332.2:c.523C>G	ENSP00000380494.2:p.Pro175Ala	2/3	YES	-	-	
S54	SNV	chr1	40366674	G	C	202	99	103	0.51	MYCL	ENSG00000116990	NON_SYNONYMOUS_CODING	ENST00000372815.1:c.523C>G	ENSP00000361902.1:p.Pro175Ala	2/2	-	-	-	
S43	SNV	chr2	160822290	T	A	681	338	343	0.504	MYO3A	ENSG00000134323	NON_SYNONYMOUS_CODING	ENST00000281043.3:c.104T>A	ENSP00000281043.2:p.Phe35Tyr	2/3	YES	-	-	
S37	SNV	chr3	38180309	C	T	547	269	278	0.508	MYD88	ENSG00000172936	NON_SYNONYMOUS_CODING	ENST00000417072.2:c.157C>T	ENSP00000401399.2:p.Arg53Trp	1/5	YES	-	-	
S37	SNV	chr3	38180309	C	T	547	269	278	0.508	MYD88	ENSG00000172936	NON_SYNONYMOUS_CODING	ENST00000495303.1:c.157C>T	ENSP00000417848.1:p.Arg53Trp	1/3	-	-	-	
S37	SNV	chr3	38180309	C	T	547	269	278	0.508	MYD88	ENSG00000172936	NON_SYNONYMOUS_CODING	ENST00000443433.2:c.157C>T	ENSP00000390565.2:p.Arg53Trp	1/4	-	-	-	
S37	SNV	chr3	38180309	C	T	547	269	278	0.508	MYD88	ENSG00000172936	NON_SYNONYMOUS_CODING	ENST00000424893.1:c.157C>T	ENSP00000389979.1:p.Arg53Trp	1/4	-	-	-	
S37	SNV	chr3	38180309	C	T	547	269	278	0.508	MYD88	ENSG00000172936	NON_SYNONYMOUS_CODING	ENST00000421516.1:c.154C>T	ENSP00000391753.1:p.Arg52Trp	1/5	-	-	-	
S37	SNV	chr3	38180309	C	T	547	269	278	0.508	MYD88	ENSG00000172936	NON_SYNONYMOUS_CODING	ENST00000396334.3:c.157C>T	ENSP00000379625.3:p.Arg53Trp	1/5	-	-	-	
33	SNV	chr16	15832439	A	G	389	203	186	0.477	MYH11	ENSG00000133392	NON_SYNONYMOUS_CODING	ENST00000452625.2:c.3125T>C	ENSP00000407821.2:p.Met1042Thr	25/43	-	0.0002	rs557463209	
33	SNV	chr16	15832439	A	G	389	203	186	0.477	MYH11	ENSG00000133392	NON_SYNONYMOUS_CODING	ENST00000300036.5:c.3104T>C	ENSP00000300036.5:p.Met1035Thr	24/41	-	0.0002	rs557463209	
33	SNV	chr16	15832439	A	G	389	203	186	0.477	MYH11	ENSG00000133392	NON_SYNONYMOUS_CODING	ENST00000396324.3:c.3125T>C	ENSP00000379616.3:p.Met1042Thr	25/42	YES	0.0002	rs557463209	
S66	SNV	chr19	8619556	G	C	644	345	299	0.464	MYO1F	ENSG00000142347	NON_SYNONYMOUS_CODING	ENST00000338257.8:c.213C>G	ENSP00000344871.6:p.Ile71Met	3/28	YES	0.0002	rs569345387,COSM1397796	
S66	SNV	chr19	8619556	G	C	644	345	299	0.464	MYO1F	ENSG00000142347	NON_SYNONYMOUS_CODING	ENST00000596675.1:c.201C>G	ENSP00000469668.1:p.Ile67Met	3/5	-	0.0002	rs569345387,COSM1397796	
62	SNV	chr8	41798416	C	T	120	56	64	0.533	MYST3	ENSG00000083168	NON_SYNONYMOUS_CODING	ENST00000265713.2:c.2983G>A	ENSP00000265713.2:p.Glu995Lys	15/17	-	-	-	
62	SNV	chr8	41798416	C	T	120	56	64	0.533	MYST3	ENSG00000083168	NON_SYNONYMOUS_CODING	ENST00000406337.1:c.2983G>A	ENSP00000385888.1:p.Glu995Lys	16/18	-	-	-	
62	SNV	chr8	41798416	C	T	120	56	64	0.533	MYST3	ENSG00000083168	NON_SYNONYMOUS_CODING	ENST00000418721.1:c.1723G>A	ENSP00000399638.1:p.Glu575Lys	8/10	-	-	-	
62	SNV	chr8	41798416	C	T	120	56	64	0.533	MYST3	ENSG00000083168	NON_SYNONYMOUS_CODING	ENST00000396930.3:c.2983G>A	ENSP00000380136.3:p.Glu995Lys	16/18	YES	-	-	
S20	SNV	chr8	41798392	G	C	950	599	351	0.37	MYST3	ENSG00000083168	NON_SYNONYMOUS_CODING	ENST00000418721.1:c.1747C>G	ENSP00000399638.1:p.Pro583Ala	8/10	-	-	-	
S20	SNV	chr8	41798392	G	C	950	599	351	0.37	MYST3	ENSG00000083168	NON_SYNONYMOUS_CODING	ENST00000396930.3:c.3007C>G	ENSP00000380136.3:p.Pro1003Ala	16/18	YES	-	-	
S20	SNV	chr8	41798392	G	C	950	599	351	0.37	MYST3	ENSG00000083168	NON_SYNONYMOUS_CODING	ENST00000265713.2:c.3007C>G	ENSP00000265713.2:p.Pro1003Ala	15/17	-	-	-	
S20	SNV	chr8	41798392	G	C	950	599	351	0.37	MYST3	ENSG00000083168	NON_SYNONYMOUS_CODING	ENST00000406337.1:c.3007C>G	ENSP00000385888.1:p.Pro1003Ala	16/18	-	-	-	
S34	INDEL	chr8	41794777	C	+A	427	301	126	0.295	MYST3	ENSG00000083168	FRAME_SHIFT	-	-	16/17	-	-	-	
S34	INDEL	chr8	41794777	C	+A	427	301	126	0.295	MYST3	ENSG00000083168	FRAME_SHIFT	-	-	17/18	-	-	-	
S34	INDEL	chr8	41794777	C	+A	427	301	126	0.295	MYST3	ENSG00000083168	FRAME_SHIFT	-	-	9/10	-	-	-	
S34	INDEL	chr8	41794777	C	+A	427	301	126	0.295	MYST3	ENSG00000083168	FRAME_SHIFT	-	-	17/18	YES	-	-	
S43	SNV	chr8	41839415	C	T	560	280	280	0.5	MYST3	ENSG00000083168	NON_SYNONYMOUS_CODING	ENST00000396930.3:c.767G>A	ENSP00000380136.3:p.Arg256Gln	5/18	YES	-	-	
S43	SNV	chr8	41839415	C	T	560	280	280	0.5	MYST3	ENSG00000083168	NON_SYNONYMOUS_CODING	ENST00000265713.2:c.767G>A	ENSP00000265713.2:p.Arg256Gln	4/17	-	-	-	
S43	SNV	chr8	41839415	C	T	560	280	280	0.5	MYST3	ENSG00000083168	NON_SYNONYMOUS_CODING	ENST00000485568.1:c.767G>A	ENSP00000430606.1:p.Arg256Gln	4/14	-	-	-	
S43	SNV	chr8	41839415	C	T	560	280	280	0.5	MYST3	ENSG00000083168	NON_SYNONYMOUS_CODING	ENST00000406337.1:c.767G>A	ENSP00000385888.1:p.Arg256Gln	5/18	-	-	-	
36	SNV	chr10	76790664	A	T	538	272	266	0.494	MYST4	ENSG00000156650	NON_SYNONYMOUS_CODING	ENST00000372711.1:c.5533A>T	ENSP00000361796.1:p.Met1845Leu	17/17	-	-	-	
36	SNV	chr10	76790664	A	T	538	272	266	0.494	MYST4	ENSG00000156650	NON_SYNONYMOUS_CODING	ENST00000372714.1:c.5206A>T	ENSP00000361799.1:p.Met1736Leu	17/17	-	-	-	
36	SNV	chr10	76790664	A	T	538	272	266	0.494	MYST4	ENSG00000156650	NON_SYNONYMOUS_CODING	ENST00000372724.1:c.5206A>T	ENSP00000361809.1:p.Met1736Leu	18/18	-	-	-	
36	SNV	chr10	76790664	A	T	538	272	266	0.494	MYST4	ENSG00000156650	NON_SYNONYMOUS_CODING	ENST00000372725.1:c.5206A>T	ENSP00000361810.1:p.Met1736Leu	17/17	-	-	-	
36	SNV	chr10	76790664	A	T	538	272	266	0.494	MYST4	ENSG00000156650	NON_SYNONYMOUS_CODING	ENST00000287239.4:c.6082A>T	ENSP00000287239.4:p.Met2028Leu	18/18	YES	-	-	
63	SNV	chr10	76789417	G	A	838	442	396	0.473	MYST4	ENSG00000156650	NON_SYNONYMOUS_CODING	ENST00000372724.1:c.3959G>A	ENSP00000361809.1:p.Arg1320His	18/18	-	0.0006	rs72803461,COSM196444	
63	SNV	chr10	76789417	G	A	838	442	396	0.473	MYST4	ENSG00000156650	NON_SYNONYMOUS_CODING	ENST00000287239.4:c.4835G>A	ENSP00000287239.4:p.Arg1612His	18/18	YES	0.0006	rs72803461,COSM196444	
63	SNV	chr10	76789417	G	A	838	442	396	0.473	MYST4	ENSG00000156650	NON_SYNONYMOUS_CODING	ENST00000372725.1:c.3959G>A	ENSP00000361810.1:p.Arg1320His	17/17	-	0.0006	rs72803461,COSM196444	
63	SNV	chr10	76789417	G	A	838	442	396	0.473	MYST4	ENSG00000156650	NON_SYNONYMOUS_CODING	ENST00000372714.1:c.3959G>A	ENSP00000361799.1:p.Arg1320His	17/17	-	0.0006	rs72803461,COSM196444	
63	SNV	chr10	76789417	G	A	838	442	396	0.473	MYST4	ENSG00000156650	NON_SYNONYMOUS_CODING	ENST00000372711.1:c.4286G>A	ENSP00000361796.1:p.Arg1429His	17/17	-	0.0006	rs72803461,COSM196444	
S22	INDEL	chr10	76781836	G	-GAGGAGGAGGAC	312	173	139	0.445	MYST4	ENSG00000156650	CODON_CHANGE_PLUS_CODON_DELETION	ENST00000372724.1:c.2344delG	ENSP00000361809.1:p.Glu782ArgfsTer40	16/18	-	-	-	
S22	INDEL	chr10	76781836	G	-GAGGAGGAGGAC	312	173	139	0.445	MYST4	ENSG00000156650	CODON_CHANGE_PLUS_CODON_DELETION	ENST00000372714.1:c.2344delG	ENSP00000361799.1:p.Glu782ArgfsTer40	15/17	-	-	-	
S22	INDEL	chr10	76781836	G	-GAGGAGGAGGAC	312	173	139	0.445	MYST4	ENSG00000156650	CODON_CHANGE_PLUS_CODON_DELETION	ENST00000372725.1:c.2344delG	ENSP00000361810.1:p.Glu782ArgfsTer40	15/17	-	-	-	
S22	INDEL	chr10	76781836	G	-GAGGAGGAGGAC	312	173	139	0.445	MYST4	ENSG00000156650	CODON_CHANGE_PLUS_CODON_DELETION	ENST00000372711.1:c.2671delG	ENSP00000361796.1:p.Glu891ArgfsTer40	15/17	-	-	-	
S25	INDEL	chr10	76781836	G	-GAGGAGGAGGAC	110	83	27	0.246	MYST4	ENSG00000156650	CODON_CHANGE_PLUS_CODON_DELETION	ENST00000287239.4:c.3220delG	ENSP00000287239.4:p.Glu1074ArgfsTer40	16/18	YES	-	-	
S25	INDEL	chr10	76781836	G	-GAGGAGGAGGAC	110	83	27	0.246	MYST4	ENSG00000156650	CODON_CHANGE_PLUS_CODON_DELETION	ENST00000287239.4:c.3220delG	ENSP00000287239.4:p.Glu1074ArgfsTer40	16/18	YES	-	-	
S25	INDEL	chr10	76781836	G	-GAGGAGGAGGAC	110	83	27	0.246	MYST4	ENSG00000156650	CODON_CHANGE_PLUS_CODON_DELETION	ENST00000372724.1:c.2344delG	ENSP00000361809.1:p.Glu782ArgfsTer40	16/18	-	-	-	
S25	INDEL	chr10	76781836	G	-GAGGAGGAGGAC	110	83	27	0.246	MYST4	ENSG00000156650	CODON_CHANGE_PLUS_CODON_DELETION	ENST00000372714.1:c.2344delG	ENSP00000361799.1:p.Glu782ArgfsTer40	15/17	-	-	-	
S25	INDEL	chr10	76781836	G	-GAGGAGGAGGAC	110	83	27	0.246	MYST4	ENSG00000156650	CODON_CHANGE_PLUS_CODON_DELETION	ENST00000372725.1:c.2344delG	ENSP00000361810.1:p.Glu782ArgfsTer40	15/17	-	-	-	
S25	INDEL	chr10	76781836	G	-GAGGAGGAGGAC	110	83	27	0.246	MYST4	ENSG00000156650	CODON_CHANGE_PLUS_CODON_DELETION	ENST00000372711.1:c.2671delG	ENSP00000361796.1:p.Glu891ArgfsTer40	15/17	-	-	-	
S55	SNV	chr10	76789417	G	A	1173	603	570	0.486	MYST4	ENSG00000156650	NON_SYNONYMOUS_CODING	ENST00000372725.1:c.3959G>A	ENSP00000361810.1:p.Arg1320His	17/17	-	0.0006	rs72803461,COSM196444	
S55	SNV	chr10	76789417	G	A	1173	603	570	0.486	MYST4	ENSG00000156650	NON_SYNONYMOUS_CODING	ENST00000372711.1:c.4286G>A	ENSP00000361796.1:p.Arg1429His	17/17	-	0.0006	rs72803461,COSM196444	
S55	SNV	chr10	76789417	G	A	1173	603	570	0.486	MYST4	ENSG00000156650	NON_SYNONYMOUS_CODING	ENST00000372724.1:c.3959G>A	ENSP00000361809.1:p.Arg1320His	18/18	-	0.0006	rs72803461,COSM196444	
S55	SNV	chr10	76789417	G	A	1173	603	570	0.486	MYST4	ENSG00000156650	NON_SYNONYMOUS_CODING	ENST00000287239.4:c.4835G>A	ENSP00000287239.4:p.Arg1612His	18/18	YES	0.0006	rs72803461,COSM196444	
S55	SNV	chr10	76789417	G	A	1173	603	570	0.486	MYST4	ENSG00000156650	NON_SYNONYMOUS_CODING	ENST00000372714.1:c.3959G>A	ENSP00000361799.1:p.Arg1320His	17/17	-	0.0006	rs72803461,COSM196444	
S62	SNV	chr10	76789417	G	A	1170	587	583	0.498	MYST4	ENSG00000156650	NON_SYNONYMOUS_CODING	ENST00000372714.1:c.3959G>A	ENSP00000361799.1:p.Arg1320His	17/17	-	0.0006	rs72803461,COSM196444	

S62	SNV	chr10	76789417	G	A	1170	587	583	0.498	MYST4	ENSG00000156650	NON_SYNONYMOUS_CODING	ENST00000372711.1:c.4286G>A	ENSP00000361796.1:p.Arg1429His	17/17	-	0.0006	rs72803461,COSM196444
S62	SNV	chr10	76789417	G	A	1170	587	583	0.498	MYST4	ENSG00000156650	NON_SYNONYMOUS_CODING	ENST00000287239.4:c.4835G>A	ENSP00000287239.4:p.Arg1612His	18/18	YES	0.0006	rs72803461,COSM196444
S62	SNV	chr10	76789417	G	A	1170	587	583	0.498	MYST4	ENSG00000156650	NON_SYNONYMOUS_CODING	ENST00000372724.1:c.3959G>A	ENSP00000361809.1:p.Arg1320His	18/18	-	0.0006	rs72803461,COSM196444
S62	SNV	chr10	76789417	G	A	1170	587	583	0.498	MYST4	ENSG00000156650	NON_SYNONYMOUS_CODING	ENST00000372725.1:c.3959G>A	ENSP00000361810.1:p.Arg1320His	17/17	-	0.0006	rs72803461,COSM196444
S65	SNV	chr10	76788793	G	A	810	428	382	0.472	MYST4	ENSG00000156650	NON_SYNONYMOUS_CODING	ENST00000287239.4:c.4211G>A	ENSP00000287239.4:p.Arg1404His	18/18	YES	-	rs563033366
S65	SNV	chr10	76788793	G	A	810	428	382	0.472	MYST4	ENSG00000156650	NON_SYNONYMOUS_CODING	ENST00000372714.1:c.3335G>A	ENSP00000361799.1:p.Arg1112His	17/17	-	-	rs563033366
S65	SNV	chr10	76788793	G	A	810	428	382	0.472	MYST4	ENSG00000156650	NON_SYNONYMOUS_CODING	ENST00000372724.1:c.3335G>A	ENSP00000361809.1:p.Arg1112His	18/18	-	-	rs563033366
S65	SNV	chr10	76788793	G	A	810	428	382	0.472	MYST4	ENSG00000156650	NON_SYNONYMOUS_CODING	ENST00000372711.1:c.3662G>A	ENSP00000361796.1:p.Arg1221His	17/17	-	-	rs563033366
S65	SNV	chr10	76788793	G	A	810	428	382	0.472	MYST4	ENSG00000156650	NON_SYNONYMOUS_CODING	ENST00000372725.1:c.3335G>A	ENSP00000361810.1:p.Arg1112His	17/17	-	-	rs563033366
25	SNV	chr12	57115079	C	T	282	145	137	0.486	NACA	ENSG00000196531	NON_SYNONYMOUS_CODING	ENST00000454682.1:c.235G>A	ENSP00000403817.1:p.Glu79Lys	3/9	-	-	-
25	SNV	chr12	57115079	C	T	282	145	137	0.486	NACA	ENSG00000196531	NON_SYNONYMOUS_CODING	ENST00000550952.1:c.235G>A	ENSP00000448035.1:p.Glu79Lys	3/11	YES	-	-
S22	SNV	chr12	57112433	A	C	174	137	37	0.213	NACA	ENSG00000196531	NON_SYNONYMOUS_CODING	ENST00000454682.1:c.2881T>G	ENSP00000403817.1:p.Trp961Gly	3/9	-	-	-
S32	SNV	chr12	57111953	G	A	140	76	64	0.457	NACA	ENSG00000196531	NON_SYNONYMOUS_CODING	ENST00000454682.1:c.3361C>T	ENSP00000403817.1:p.Pro1121Ser	3/9	-	0.0002	rs544182420
S37	SNV	chr12	57114484	G	C	759	391	368	0.485	NACA	ENSG00000196531	NON_SYNONYMOUS_CODING	ENST00000454682.1:c.830C>G	ENSP00000403817.1:p.Ser277Cys	3/9	-	0.0004	rs543527270
S37	SNV	chr12	57114484	G	C	759	391	368	0.485	NACA	ENSG00000196531	NON_SYNONYMOUS_CODING	ENST00000550952.1:c.830C>G	ENSP00000448035.1:p.Ser277Cys	3/11	YES	0.0004	rs543527270
20	SNV	chr8	90965695	G	C	255	137	118	0.463	NBN	ENSG00000104320	NON_SYNONYMOUS_CODING	ENST00000265433.3:c.1622C>G	ENSP00000265433.3:p.Ala541Gly	11/16	YES	-	-
20	SNV	chr8	90965695	G	C	255	137	118	0.463	NBN	ENSG00000104320	NON_SYNONYMOUS_CODING	ENST00000469330.1:c.1376C>G	ENSP00000386924.1:p.Ala459Gly	11/16	-	-	-
63	SNV	chr2	24952682	C	T	426	208	218	0.512	NCOA1	ENSG00000084676	STOP_GAINED	ENST00000406961.1:c.3199C>T	ENSP00000385216.1:p.Gln1067Ter	17/23	YES	-	-
63	SNV	chr2	24952682	C	T	426	208	218	0.512	NCOA1	ENSG00000084676	STOP_GAINED	ENST00000405141.1:c.3199C>T	ENSP00000385097.1:p.Gln1067Ter	18/25	-	-	-
63	SNV	chr2	24952682	C	T	426	208	218	0.512	NCOA1	ENSG00000084676	STOP_GAINED	ENST00000395856.3:c.3199C>T	ENSP00000379197.3:p.Gln1067Ter	15/21	-	-	-
63	SNV	chr2	24952682	C	T	426	208	218	0.512	NCOA1	ENSG00000084676	STOP_GAINED	ENST00000348332.3:c.3199C>T	ENSP00000320940.5:p.Gln1067Ter	15/21	-	-	-
63	SNV	chr2	24952682	C	T	426	208	218	0.512	NCOA1	ENSG00000084676	STOP_GAINED	ENST00000538539.1:c.3199C>T	ENSP00000444039.1:p.Gln1067Ter	16/23	-	-	-
63	SNV	chr2	24952682	C	T	426	208	218	0.512	NCOA1	ENSG00000084676	STOP_GAINED	ENST00000288599.5:c.3199C>T	ENSP00000288599.5:p.Gln1067Ter	15/22	-	-	-
63	SNV	chr2	24952682	C	T	426	208	218	0.512	NCOA1	ENSG00000084676	STOP_GAINED	ENST00000407230.1:c.2746C>T	ENSP00000385195.1:p.Gln916Ter	15/22	-	-	-
S53	SNV	chr2	24929784	C	T	750	385	365	0.487	NCOA1	ENSG00000084676	NON_SYNONYMOUS_CODING	ENST00000395856.3:c.1445C>G	ENSP00000379197.3:p.Ser482Cys	11/21	-	-	rs555210413
S53	SNV	chr2	24929784	C	G	750	385	365	0.487	NCOA1	ENSG00000084676	NON_SYNONYMOUS_CODING	ENST00000348332.3:c.1445C>G	ENSP00000320940.5:p.Ser482Cys	11/21	-	-	rs555210413
S53	SNV	chr2	24929784	C	G	750	385	365	0.487	NCOA1	ENSG00000084676	NON_SYNONYMOUS_CODING	ENST00000406961.1:c.1445C>G	ENSP00000385216.1:p.Ser482Cys	13/23	YES	-	rs555210413
S53	SNV	chr2	24929784	C	G	750	385	365	0.487	NCOA1	ENSG00000084676	NON_SYNONYMOUS_CODING	ENST00000288599.5:c.1445C>G	ENSP00000288599.5:p.Ser482Cys	11/22	-	-	rs555210413
S53	SNV	chr2	24929784	C	G	750	385	365	0.487	NCOA1	ENSG00000084676	NON_SYNONYMOUS_CODING	ENST00000538539.1:c.1445C>G	ENSP00000444039.1:p.Ser482Cys	12/23	-	-	rs555210413
S53	SNV	chr2	24929784	C	G	750	385	365	0.487	NCOA1	ENSG00000084676	NON_SYNONYMOUS_CODING	ENST00000405141.1:c.1445C>G	ENSP00000385097.1:p.Ser482Cys	14/25	-	-	rs555210413
S53	SNV	chr2	24929784	C	G	750	385	365	0.487	NCOA1	ENSG00000084676	NON_SYNONYMOUS_CODING	ENST00000407230.1:c.992C>G	ENSP00000385195.1:p.Ser331Cys	11/22	-	-	rs555210413
40	SNV	chr10	21101814	A	G	39	20	19	0.487	NEBL	ENSG00000078114	NON_SYNONYMOUS_CODING	ENST00000377122.4:c.2402T>C	ENSP00000366326.4:p.Val801Ala	24/28	YES	-	-
40	SNV	chr10	21101814	A	G	39	20	19	0.487	NEBL	ENSG00000078114	NON_SYNONYMOUS_CODING	ENST00000377159.4:c.311T>C	ENSP00000366364.4:p.Val104Ala	4/6	-	-	-
40	SNV	chr10	21101814	A	G	39	20	19	0.487	NEBL	ENSG00000078114	NON_SYNONYMOUS_CODING	ENST0000041778.2:c.413T>C	ENSP00000393896.2:p.Val138Ala	5/7	-	-	-
62	SNV	chr10	21358886	G	A	156	72	84	0.538	NEBL	ENSG00000078114	NON_SYNONYMOUS_CODING	ENST00000377159.4:c.59C>T	ENSP00000366364.4:p.Pro20Leu	1/6	-	0.0006	rs572181068
28	INDEL	chr17	29553477	A	+C	319	190	129	0.404	NF1	ENSG00000196712	FRAME_SHIFT	-	-	10/50	-	-	COSM36864
28	INDEL	chr17	29553477	A	+C	319	190	129	0.404	NF1	ENSG00000196712	FRAME_SHIFT	-	-	18/58	YES	-	COSM36864
28	INDEL	chr17	29553477	A	+C	319	190	129	0.404	NF1	ENSG00000196712	FRAME_SHIFT	-	-	18/57	-	-	COSM36864
36	SNV	chr17	29554590	T	A	398	215	183	0.46	NF1	ENSG00000196712	NON_SYNONYMOUS_CODING	ENST00000356175.3:c.2375T>A	ENSP00000348498.3:p.Leu792His	20/57	-	-	COSM560397,COSM560396
36	SNV	chr17	29554590	T	A	398	215	183	0.46	NF1	ENSG00000196712	NON_SYNONYMOUS_CODING	ENST00000456735.2:c.1373T>A	ENSP00000389907.2:p.Leu458His	12/50	-	-	COSM560397,COSM560396
36	SNV	chr17	29554590	T	A	398	215	183	0.46	NF1	ENSG00000196712	NON_SYNONYMOUS_CODING	ENST00000358273.4:c.2375T>A	ENSP00000351015.4:p.Leu792His	20/58	YES	-	COSM560397,COSM560396
48	SNV	chr17	29560206	C	T	328	164	164	0.5	NF1	ENSG00000196712	NON_SYNONYMOUS_CODING	ENST00000456735.2:c.2681C>T	ENSP00000389907.2:p.Ala894Val	19/50	-	-	-
48	SNV	chr17	29560206	C	T	328	164	164	0.5	NF1	ENSG00000196712	NON_SYNONYMOUS_CODING	ENST00000358273.4:c.3683C>T	ENSP00000351015.4:p.Ala1228Val	27/58	YES	-	-
48	SNV	chr17	29560206	C	T	328	164	164	0.5	NF1	ENSG00000196712	NON_SYNONYMOUS_CODING	ENST00000356175.3:c.3683C>T	ENSP00000348498.3:p.Ala1228Val	27/57	-	-	-
S10	INDEL	chr17	29553477	A	+C	811	503	308	0.378	NF1	ENSG00000196712	FRAME_SHIFT	-	-	18/57	-	-	COSM36864
S10	INDEL	chr17	29553477	A	+C	811	503	308	0.378	NF1	ENSG00000196712	FRAME_SHIFT	-	-	18/58	YES	-	COSM36864
S10	SNV	chr17	29562979	T	A	561	434	127	0.226	NF1	ENSG00000196712	STOP_GAINED	ENST00000356175.3:c.3914T>A	ENSP00000348498.3:p.Leu1305Ter	29/57	-	-	-
S10	SNV	chr17	29562979	T	A	561	434	127	0.226	NF1	ENSG00000196712	STOP_GAINED	ENST00000456735.2:c.2912T>A	ENSP00000389907.2:p.Leu971Ter	21/50	-	-	-
S10	SNV	chr17	29562979	T	A	561	434	127	0.226	NF1	ENSG00000196712	STOP_GAINED	ENST00000358273.4:c.3914T>A	ENSP00000351015.4:p.Leu1305Ter	29/58	YES	-	-
S10	INDEL	chr17	29553477	A	+C	811	503	308	0.378	NF1	ENSG00000196712	FRAME_SHIFT	-	-	10/50	-	-	COSM36864
S29	SNV	chr22	30032840	T	C	342	175	167	0.488	NF2	ENSG00000186575	NON_SYNONYMOUS_CODING	ENST00000361452.4:c.215T>C	ENSP00000354897.4:p.Val72Ala	2/16	-	-	-
S29	SNV	chr22	30032840	T	C	342	175	167	0.488	NF2	ENSG00000186575	NON_SYNONYMOUS_CODING	ENST00000338641.4:c.215T>C	ENSP00000344666.4:p.Val72Ala	2/16	YES	-	-
S29	SNV	chr22	30032840	T	C	342	175	167	0.488	NF2	ENSG00000186575	NON_SYNONYMOUS_CODING	ENST00000403435.1:c.215T>C	ENSP00000384029.1:p.Val72Ala	2/17	-	-	-
S29	SNV	chr22	30032840	T	C	342	175	167	0.488	NF2	ENSG00000186575	NON_SYNONYMOUS_CODING	ENST00000413209.2:c.215T>C	ENSP00000409921.2:p.Val72Ala	2/5	-	-	-
S29	SNV	chr22	30032840	T	C	342	175	167	0.488	NF2	ENSG00000186575	NON_SYNONYMOUS_CODING	ENST00000403999.3:c.215T>C	ENSP00000384797.3:p.Val72Ala	2/16	-	-	-
S29	SNV	chr22	30032840	T	C	342	175	167	0.488	NF2	ENSG00000186575	NON_SYNONYMOUS_CODING	ENST00000397789.3:c.215T>C	ENSP00000380891.3:p.Val72Ala	2/17	-	-	-
S29	SNV	chr22	30032840	T	C	342	175	167	0.488	NF2	ENSG00000186575	NON_SYNONYMOUS_CODING	ENST00000361166.4:c.215T>C	ENSP00000354529.4:p.Val72Ala	2/17	-	-	-
S42	SNV	chr2	178097244	A	G	519	264	255	0.491	NFE2L2	ENSG00000116044	NON_SYNONYMOUS_CODING	ENST00000421929.1:c.422T>C	ENSP00000412191.1:p.Ile141Thr	4/5	-	-	-
S42	SNV	chr2	178097244	A	G	519	264	255	0.491	NFE2L2	ENSG00000116044	NON_SYNONYMOUS_CODING	ENST00000397062.3:c.470T>C	ENSP00000380252.3:p.Ile157Thr	4/5	YES	-	-
S42	SNV	chr2	178097244	A	G	519	264	255	0.491	NFE2L2	ENSG00000116044	NON_SYNONYMOUS_CODING	ENST00000586532.1:c.467T>C	ENSP00000464920.1:p.Ile156Thr	5/6	-	-	-
S42	SNV	chr2	178097244	A	G	519	264	255	0.491	NFE2L2	ENSG00000116044	NON_SYNONYMOUS_CODING	ENST00000461575.2:c.401T>C	ENSP00000411575.2:p.Ile134Thr	4/5	-	-	-
S42	SNV	chr2	178097244	A	G	519	264	255	0.491	NFE2L2	ENSG00000116044	NON_SYNONYMOUS_CODING	ENST00000397063.4:c.422T>C	ENSP00000380253.4:p.Ile141Thr	4/5	-	-	-
S42	SNV	chr2	178097244	A	G	519	264	255	0.491	NFE2L2	ENSG00000116044	NON_SYNONYMOUS_CODING	ENST00000588123.1:c.422T>C	ENSP00000468089.1:p.Ile141Thr	5/5	-	-	-
S42	SNV	chr2	178097244	A	G	519	264	255	0.491	NFE2L2	ENSG00000116044	NON_SYNONYMOUS_CODING	ENST00000448782.1:c.422T>C	ENSP00000400073.1:p.Ile141Thr	4/5	-	-	-
S42	SNV	chr2	178097244	A	G	519	264	255	0.491	NFE2L2	ENSG00000116044	NON_SYNONYMOUS_CODING	ENST00000464747.1:c.422T>C	ENSP00000467401.1:p.Ile141Thr	7/8	-	-	-
S54	SNV	chr2	178129270	G	A	199	89	110	0.553	NFE2L2	ENSG00000116044	NON_SYNONYMOUS_CODING	ENST00000397062.3:c.35C>T	ENSP00000380252.3:p.Pro12Leu	1/5	YES	-	-
S54	SNV	chr2	178129270	G	A	199	89	110	0.553	NFE2L2	ENSG00000116044	NON_SYNONYMOUS_CODING	ENST00000430047.1:c.35C>T	ENSP00000391291.1:p.Pro12Leu	1/3	-	-	-
33	SNV	chr14	51224492	T	C	677	368	309	0.456	NIN	ENSG00000100503	NON_SYNONYMOUS_CODING	ENST00000245441.5:c.3256A>G	ENSP00000245441.5:p.Thr1086Ala	18/31	-	-	-
33	SNV	chr14	51224492	T	C	677	368	309	0.456	NIN	ENSG00000100503	NON_SYNONYMOUS_CODING	ENST00000382041.3:c.3256A>G	ENSP00000371472.3:p.Thr1086Ala	18/30	YES	-	-
33	SNV	chr14	51224492	T	C	677	368	309	0.456	NIN	ENSG00000100503	NON_SYNONYMOUS_CODING	ENST00000324330.9:c.3256A>G	ENSP00000324210.9:p.Thr1086Ala	18/31	-	-	-
33	SNV	chr14	51224492	T	C	677	368	309	0.456	NIN	ENSG00000100503	NON_SYNONYMOUS_CODING	ENST00000453196.1:c.3256A>G	ENSP00000412391.1:p.Thr1086Ala	18/30	-	-	-
S37	SNV	chr14	51224173	T	G	707	346	361	0.511	NIN	ENSG00000100503	NON_SYNONYMOUS_CODING	ENST00000453196.1:c.3575A>C	ENSP00000412391.1:p.Glu1192Ala	18/30	-	-	-
S37	SNV	chr14	51224173	T	G	707	346	361	0.511	NIN	ENSG00000100503	NON_SYNONYMOUS_CODING	ENST00000324330.9:c.3575A>C	ENSP00000324210.9:p.Glu1192Ala	18/31	-	-	-
S37	SNV	chr14	51224173	T	G	707	346	361	0.511	NIN	ENSG00000100503	NON_SYNONYMOUS_CODING	ENST00000382041.3:c.3575A>C	ENSP00000371472.3:p.Glu1192Ala	18/30	YES	-	-
S37	SNV	chr14	51224173	T	G	707	346	361	0.511	NIN	ENSG00000100503	NON_SYNONYMOUS_CODING	ENST00000389869.3:c.2046A>C	ENSP00000374519.3:p.Glu683Asp	6/18	-	-	-
S37	SNV	chr14	51224173	T	G	707	346	361	0.511	NIN	ENSG00000100503	NON_SYNONYMOUS_CODING	ENST00000530853.1:c.2046A>C	ENSP00000433716.1:p.Glu683Asp	6/18	-	-	-
S37	SNV	chr14	51224173	T	G	707	346	361	0.511	NIN	ENSG00000100503	NON_SYNONYMOUS_CODING	ENST00000530997.2:c.3575A>C	ENSP00000436092.2:p.Glu1192Ala	16/29	-	-	-
S37	SNV	chr14	51224173	T	G	707	346	361	0.511	NIN	ENSG00000100503	NON_SYNONYMOUS_CODING	ENST00000245441.5:c.3575A>C	ENSP00000245441.5:p.Glu1192Ala	18/31	-	-	-
34	SNV	chr14	36988763	T	G	224	112	112	0.5	NKX2-1	ENSG00000136352	UTR_5_PRIME	ENST00000498187.2:c.-201A>C	-	1/2	-	-	rs561607086
34	SNV	chr14	36988763	T	G	224	112	112	0.5	NKX2-1	ENSG00000136352	UTR_5_PRIME	ENST00000522719.2:c.-104A>C	-	2/4	-	-	rs561607086
19	SNV	chr9	139393401	C	T	695	377	318	0.458	NOTCH1	ENSG00000148400	NON_SYNONYMOUS_CODING	ENST00000277541.6:c.6130G>A	ENSP00000277541.6:p.Ala2044Thr	33/34	YES	0.0002	rs544856644
25	SNV	chr9	139405180	C	T	124	73	51	0.411	NOTCH1	ENSG00000148400	NON_SYNONYMOUS_CODING	ENST00000277541.6:c.2665G>A	ENSP00000277541.6:p.Gly889Ser	17/34	YES	0.0002	rs557590721
32	SNV	chr1	120506232	T	C	125	96	29	0.232	NOTCH2	ENSG00000134250	NON_SYNONYMOUS_CODING	ENST00000256646.2:c.1880A>G	ENSP00000256646.2:p.Asn627Ser	11/34	-	-	COSM3934035
19	INDEL	chr5	170837543	C	+TCTG	37	26	11	0.297	NPM1	ENSG00000181163	FRAME_SHIFT	-	-	11/11	YES	-	-
19	INDEL	chr5	170837543	C	+TCTG	37	26	11	0.297	NPM1	ENSG00000181163	FRAME_SHIFT	-	-	12/12	-	-	-
19	INDEL	chr5	170837543	C	+TCTG	37	26	11	0.297	NPM1	ENSG00000181163	FRAME_SHIFT	-	-	10/10	-	-	-
33	INDEL	chr5	170837545	C	+TGCA	103	62	41	0.398	NPM1	ENSG00000181163	FRAME_SHIFT	-	-	10/10	-	-	COSM36379
33	INDEL	chr5	170837545	C	+TGCA	103	62	41	0.398	NPM1	ENSG00000181163	FRAME_SHIFT	-	-	12/12	-	-	COSM36379
33	INDEL	chr5	170837545	C	+TGCA	103	62	41	0.398	NPM1	ENSG00000181163	FRAME_SHIFT	-	-	11/11	YES	-	COSM36379
42	INDEL	chr5	170837543	C	+TCTG	159	100	59	0.371	NPM1	ENSG00000181163	FRAME_SHIFT	-	-	10/10	-	-	-
42	INDEL	chr5	170837543	C	+TCTG	159	100	59	0.371	NPM1	ENSG00000181163	FRAME_SHIFT	-	-	11/11	YES	-	-
42	INDEL	chr5	170837543	C	+TCTG	159	100	59	0.371	NPM1	ENSG00000181163	FRAME_SHIFT	-	-	12/12	-	-	-
46	INDEL	chr5	170837543	C	+TCTG	143	94	49	0.34	NPM1	ENSG00000181163	FRAME_SHIFT	-	-	11/11	YES	-	-
46	INDEL	chr5	170837543	C	+TCTG	143	94	49	0.34	NPM1	ENSG00000181163	FRAME_SHIFT	-	-	10/10	-	-	-
46	INDEL	chr5	170837543	C	+TCTG	143	94	49	0.34	NPM1	ENSG00000181163	FRAME_SHIFT	-	-	12/12	-	-	-
59	INDEL	chr5	170837545	C	+TGCA	114	66	48	0.421	NPM1	ENSG00000181163	FRAME_SHIFT	-	-	12/12	-	-	COSM36379
59	INDEL	chr5	170837545	C	+TGCA	114	66	48	0.421	NPM1	ENSG00000181163	FRAME_SHIFT	-	-	11/11	YES	-	COSM36379
59	INDEL	chr5	170837545	C	+TGCA	114	66	48	0.421	NPM1	ENSG00000181163	FRAME_SHIFT	-	-	10/10	-	-	COSM36379
7	INDEL	chr5	170837543	C	+TCTG	92	66	26	0.283	NPM1	ENSG00000181163	FRAME_SHIFT	-	-	10/10	-	-	-

7	INDEL	chr5	170837543	C	+TCTG	92	66	26	0.283	NPM1	ENSG00000181163	FRAME_SHIFT	-	-	11/11	YES	-	-	
7	INDEL	chr5	170837543	C	+TCTG	92	66	26	0.283	NPM1	ENSG00000181163	FRAME_SHIFT	-	-	12/12	-	-	-	
S02	INDEL	chr5	170837544	T	+CTGC	66	39	27	0.409	NPM1	ENSG00000181163	FRAME_SHIFT	-	-	10/10	-	-	-	
S02	INDEL	chr5	170837544	T	+CTGC	66	39	27	0.409	NPM1	ENSG00000181163	FRAME_SHIFT	-	-	12/12	-	-	-	
S02	INDEL	chr5	170837544	T	+CTGC	66	39	27	0.409	NPM1	ENSG00000181163	FRAME_SHIFT	-	-	11/11	YES	-	-	
S06	INDEL	chr5	170837543	C	+TCTG	88	44	44	0.484	NPM1	ENSG00000181163	FRAME_SHIFT	-	-	10/10	-	-	-	
S06	INDEL	chr5	170837543	C	+TCTG	88	44	44	0.484	NPM1	ENSG00000181163	FRAME_SHIFT	-	-	11/11	YES	-	-	
S06	INDEL	chr5	170837543	C	+TCTG	88	44	44	0.484	NPM1	ENSG00000181163	FRAME_SHIFT	-	-	12/12	-	-	-	
S32	INDEL	chr5	170837543	C	+TCTG	133	75	58	0.409	NPM1	ENSG00000181163	FRAME_SHIFT	-	-	10/10	-	-	-	
S32	INDEL	chr5	170837543	C	+TCTG	133	75	58	0.409	NPM1	ENSG00000181163	FRAME_SHIFT	-	-	11/11	YES	-	-	
S32	INDEL	chr5	170837543	C	+TCTG	133	75	58	0.409	NPM1	ENSG00000181163	FRAME_SHIFT	-	-	12/12	-	-	-	
S45	INDEL	chr5	170837543	C	+TCTG	133	83	50	0.37	NPM1	ENSG00000181163	FRAME_SHIFT	-	-	12/12	-	-	-	
S45	INDEL	chr5	170837543	C	+TCTG	133	83	50	0.37	NPM1	ENSG00000181163	FRAME_SHIFT	-	-	11/11	YES	-	-	
S45	INDEL	chr5	170837543	C	+TCTG	133	83	50	0.37	NPM1	ENSG00000181163	FRAME_SHIFT	-	-	10/10	-	-	-	
S47	INDEL	chr5	170837544	T	+CTGC	103	63	40	0.381	NPM1	ENSG00000181163	FRAME_SHIFT	-	-	11/11	YES	-	-	
S47	INDEL	chr5	170837544	T	+CTGC	103	63	40	0.381	NPM1	ENSG00000181163	FRAME_SHIFT	-	-	12/12	-	-	-	
S47	INDEL	chr5	170837544	T	+CTGC	103	63	40	0.381	NPM1	ENSG00000181163	FRAME_SHIFT	-	-	10/10	-	-	-	
S52	INDEL	chr5	170837543	C	+TCTG	165	118	47	0.278	NPM1	ENSG00000181163	FRAME_SHIFT	-	-	10/10	-	-	-	
S52	INDEL	chr5	170837543	C	+TCTG	165	118	47	0.278	NPM1	ENSG00000181163	FRAME_SHIFT	-	-	11/11	YES	-	-	
S52	INDEL	chr5	170837543	C	+TCTG	165	118	47	0.278	NPM1	ENSG00000181163	FRAME_SHIFT	-	-	12/12	-	-	-	
S57	INDEL	chr5	170837543	C	+TCTG	203	109	94	0.456	NPM1	ENSG00000181163	FRAME_SHIFT	-	-	12/12	-	-	-	
S57	INDEL	chr5	170837543	C	+TCTG	203	109	94	0.456	NPM1	ENSG00000181163	FRAME_SHIFT	-	-	11/11	YES	-	-	
S57	INDEL	chr5	170837543	C	+TCTG	203	109	94	0.456	NPM1	ENSG00000181163	FRAME_SHIFT	-	-	10/10	-	-	-	
S65	INDEL	chr5	170837543	C	+TCTG	180	122	58	0.319	NPM1	ENSG00000181163	FRAME_SHIFT	-	-	10/10	-	-	-	
S65	INDEL	chr5	170837543	C	+TCTG	180	122	58	0.319	NPM1	ENSG00000181163	FRAME_SHIFT	-	-	11/11	YES	-	-	
S65	INDEL	chr5	170837543	C	+TCTG	180	122	58	0.319	NPM1	ENSG00000181163	FRAME_SHIFT	-	-	12/12	-	-	-	
S66	INDEL	chr5	170837543	C	+TCTG	136	76	60	0.432	NPM1	ENSG00000181163	FRAME_SHIFT	-	-	10/10	-	-	-	
S66	INDEL	chr5	170837543	C	+TCTG	136	76	60	0.432	NPM1	ENSG00000181163	FRAME_SHIFT	-	-	12/12	-	-	-	
S66	INDEL	chr5	170837543	C	+TCTG	136	76	60	0.432	NPM1	ENSG00000181163	FRAME_SHIFT	-	-	11/11	YES	-	-	
32	SNV	chr9	102626046	T	A	128	80	48	0.375	NR4A3	ENSG00000119508	NON_SYNONYMOUS_CODING	ENST00000395097.2:c.1778T>A	ENSP00000378531.2:p.Ile593Asn	8/8	-	-	-	
32	SNV	chr9	102626046	T	A	128	80	48	0.375	NR4A3	ENSG00000119508	NON_SYNONYMOUS_CODING	ENST00000330847.1:c.1811T>A	ENSP00000333122.1:p.Ile604Asn	7/7	YES	-	-	
S20	INDEL	chr9	102590524	G	-CAA	717	377	340	0.474	NR4A3	ENSG00000119508	CODON_CHANGE_PLUS_CODON_DELETION	ENST00000338488.4:c.201delC	ENSP00000340301.4:p.Ser67ArgfsTer17	3/5	-	-	-	
S20	INDEL	chr9	102590524	G	-CAA	717	377	340	0.474	NR4A3	ENSG00000119508	CODON_CHANGE_PLUS_CODON_DELETION	ENST00000395097.2:c.201delC	ENSP00000378531.2:p.Ser67ArgfsTer17	3/8	-	-	-	
S20	INDEL	chr9	102590524	G	-CAA	717	377	340	0.474	NR4A3	ENSG00000119508	CODON_CHANGE_PLUS_CODON_DELETION	ENST00000330847.1:c.234delC	ENSP00000333122.1:p.Ser78ArgfsTer17	2/7	YES	-	-	
42	SNV	chr1	115256529	T	C	193	131	62	0.321	NRAS	ENSG00000213281	NON_SYNONYMOUS_CODING	ENST00000369535.4:c.182A>G	ENSP00000358548.4:p.Gln61Arg	3/7	YES	-	COSM584,COSM583,COSM582	
S18	SNV	chr1	115256535	G	C	485	280	205	0.423	NRAS	ENSG00000213281	NON_SYNONYMOUS_CODING	ENST00000369535.4:c.176C>G	ENSP00000358548.4:p.Ala59Gly	3/7	YES	-	COSM253327	
S25	SNV	chr1	115258747	C	T	50	31	19	0.38	NRAS	ENSG00000213281	NON_SYNONYMOUS_CODING	ENST00000369535.4:c.35G>A	ENSP00000358548.4:p.Gly12Asp	2/7	YES	-	COSM565,COSM564,COSM566	
S46	SNV	chr1	115256530	G	T	498	339	159	0.319	NRAS	ENSG00000213281	NON_SYNONYMOUS_CODING	ENST00000369535.4:c.181C>A	ENSP00000358548.4:p.Gln61Lys	3/7	YES	-	COSM580,COSM581	
38	SNV	chr5	176631227	T	A	747	373	374	0.501	NSD1	ENSG00000165671	NON_SYNONYMOUS_CODING	ENST00000354179.4:c.3631>A	ENSP00000346111.4:p.His1211Gln	5/24	-	-	-	
38	SNV	chr5	176631227	T	A	747	373	374	0.501	NSD1	ENSG00000165671	NON_SYNONYMOUS_CODING	ENST00000510954.1:c.3631>A	ENSP00000423982.1:p.His1211Gln	5/5	-	-	-	
38	SNV	chr5	176631227	T	A	747	373	374	0.501	NSD1	ENSG00000165671	NON_SYNONYMOUS_CODING	ENST00000508896.1:c.3631>A	ENSP00000423372.1:p.His1211Gln	5/6	-	-	-	
38	SNV	chr5	176631227	T	A	747	373	374	0.501	NSD1	ENSG00000165671	NON_SYNONYMOUS_CODING	ENST00000347982.4:c.3631>A	ENSP00000343209.4:p.His1211Gln	5/24	-	-	-	
38	SNV	chr5	176631227	T	A	747	373	374	0.501	NSD1	ENSG00000165671	NON_SYNONYMOUS_CODING	ENST00000439151.2:c.1170T>A	ENSP00000395929.2:p.His390Gln	4/23	YES	-	-	
48	SNV	chr5	176709485	A	G	307	191	116	0.378	NSD1	ENSG00000165671	NON_SYNONYMOUS_CODING	ENST00000347982.4:c.5105A>G	ENSP00000343209.4:p.Tyr1702Cys	20/24	-	-	COSM1166637,COSM1166638	
48	SNV	chr5	176709485	A	G	307	191	116	0.378	NSD1	ENSG00000165671	NON_SYNONYMOUS_CODING	ENST00000439151.2:c.5912A>G	ENSP00000395929.2:p.Tyr1971Cys	19/23	YES	-	COSM1166637,COSM1166638	
48	SNV	chr5	176709485	A	G	307	191	116	0.378	NSD1	ENSG00000165671	NON_SYNONYMOUS_CODING	ENST00000354179.4:c.5105A>G	ENSP00000346111.4:p.Tyr1702Cys	20/24	-	-	COSM1166637,COSM1166638	
48	SNV	chr5	176709485	A	G	307	191	116	0.378	NSD1	ENSG00000165671	NON_SYNONYMOUS_CODING	ENST00000361032.4:c.5603A>G	ENSP00000354310.4:p.Tyr1868Cys	16/20	-	-	COSM1166637,COSM1166638	
59	SNV	chr5	176709485	A	G	111	62	49	0.441	NSD1	ENSG00000165671	NON_SYNONYMOUS_CODING	ENST00000361032.4:c.5603A>G	ENSP00000354310.4:p.Tyr1868Cys	16/20	-	-	COSM1166637,COSM1166638	
59	SNV	chr5	176709485	A	G	111	62	49	0.441	NSD1	ENSG00000165671	NON_SYNONYMOUS_CODING	ENST00000439151.2:c.5912A>G	ENSP00000395929.2:p.Tyr1971Cys	19/23	YES	-	COSM1166637,COSM1166638	
59	SNV	chr5	176709485	A	G	111	62	49	0.441	NSD1	ENSG00000165671	NON_SYNONYMOUS_CODING	ENST00000354179.4:c.5105A>G	ENSP00000346111.4:p.Tyr1702Cys	20/24	-	-	COSM1166637,COSM1166638	
59	SNV	chr5	176709485	A	G	111	62	49	0.441	NSD1	ENSG00000165671	NON_SYNONYMOUS_CODING	ENST00000347982.4:c.5105A>G	ENSP00000343209.4:p.Tyr1702Cys	20/24	-	-	COSM1166637,COSM1166638	
31	INDEL	chr9	134107752	A	-GT	269	168	101	0.375	NUP214	ENSG00000126883	UTR_3_PRIME	ENST00000498610.1:c.*38delG	-	3/3	-	-	-	
37	SNV	chr9	134072846	T	A	106	55	51	0.481	NUP214	ENSG00000126883	NON_SYNONYMOUS_CODING	ENST00000411637.2:c.3935T>A	ENSP00000396576.2:p.Phe1312Tyr	29/36	-	-	-	
37	SNV	chr9	134072846	T	A	106	55	51	0.481	NUP214	ENSG00000126883	NON_SYNONYMOUS_CODING	ENST00000451030.1:c.3968T>A	ENSP00000404950.1:p.Phe1323Tyr	29/36	-	-	-	
37	SNV	chr9	134072846	T	A	106	55	51	0.481	NUP214	ENSG00000126883	NON_SYNONYMOUS_CODING	ENST00000359428.5:c.3965T>A	ENSP00000352400.5:p.Phe1322Tyr	29/36	YES	-	-	
37	SNV	chr9	134072846	T	A	106	55	51	0.481	NUP214	ENSG00000126883	NON_SYNONYMOUS_CODING	ENST00000531600.1:c.296T>A	ENSP00000435364.1:p.Phe99Tyr	2/2	-	-	-	
37	SNV	chr9	134072846	T	A	106	55	51	0.481	NUP214	ENSG00000126883	NON_SYNONYMOUS_CODING	ENST00000483497.2:c.443T>A	ENSP00000436793.1:p.Phe148Tyr	4/11	-	-	-	
40	SNV	chr9	134022967	C	T	94	46	48	0.511	NUP214	ENSG00000126883	NON_SYNONYMOUS_CODING	ENST00000411637.2:c.2006C>T	ENSP00000396576.2:p.Pro669Leu	14/36	-	-	-	
40	SNV	chr9	134022967	C	T	94	46	48	0.511	NUP214	ENSG00000126883	NON_SYNONYMOUS_CODING	ENST00000451030.1:c.2039C>T	ENSP00000404014.1:p.Pro680Leu	14/36	-	-	-	
40	SNV	chr9	134022967	C	T	94	46	48	0.511	NUP214	ENSG00000126883	NON_SYNONYMOUS_CODING	ENST00000359428.5:c.2036C>T	ENSP00000352400.5:p.Pro679Leu	14/36	YES	-	-	
S05	SNV	chr11	3726535	C	T	512	269	243	0.475	NUP98	ENSG00000110713	NON_SYNONYMOUS_CODING	ENST00000359171.4:c.2977G>A	ENSP00000352091.4:p.Asp993Asn	22/34	-	-	-	
S05	SNV	chr11	3726535	C	T	512	269	243	0.475	NUP98	ENSG00000110713	NON_SYNONYMOUS_CODING	ENST00000329801.1:c.143G>A	ENSP00000413146.1:p.Arg48Asn	2/13	-	-	-	
S05	SNV	chr11	3726535	C	T	512	269	243	0.475	NUP98	ENSG00000110713	NON_SYNONYMOUS_CODING	ENST00000324932.7:c.2977G>A	ENSP00000316032.7:p.Asp993Asn	22/33	YES	-	-	
S05	SNV	chr11	3726535	C	T	512	269	243	0.475	NUP98	ENSG00000110713	NON_SYNONYMOUS_CODING	ENST00000355260.3:c.2977G>A	ENSP00000347404.3:p.Asp993Asn	22/32	-	-	-	
S64	SNV	chr11	3784267	T	A	501	258	243	0.485	NUP98	ENSG00000110713	SPLICE_SITE_REGION	ENST00000355260.3:c.951A>T	ENST00000355260.3:c.951A>T(p.%3D)	9/32	-	-	-	
S64	SNV	chr11	3784267	T	A	501	258	243	0.485	NUP98	ENSG00000110713	SPLICE_SITE_REGION	ENST00000397007.4:c.951A>T	ENST00000397007.4:c.951A>T(p.%3D)	9/20	-	-	-	
S64	SNV	chr11	3784267	T	A	501	258	243	0.485	NUP98	ENSG00000110713	SPLICE_SITE_REGION	ENST00000324932.7:c.951A>T	ENST00000324932.7:c.951A>T(p.%3D)	9/33	YES	-	-	
S64	SNV	chr11	3784267	T	A	501	258	243	0.485	NUP98	ENSG00000110713	SPLICE_SITE_REGION	ENST00000359171.4:c.951A>T	ENST00000359171.4:c.951A>T(p.%3D)	9/34	-	-	-	
S64	SNV	chr11	3784267	T	A	501	258	243	0.485	NUP98	ENSG00000110713	SPLICE_SITE_REGION	ENST00000397004.4:c.951A>T	ENST00000397004.4:c.951A>T(p.%3D)	9/20	-	-	-	
S14	SNV	chr15	34640808	C	T	369	186	183	0.496	NUTM1	ENSG00000184507	NON_SYNONYMOUS_CODING	ENST00000537011.1:c.739C>T	ENSP00000444896.1:p.Arg247Cys	3/8	-	-	-	
S14	SNV	chr15	34640808	C	T	369	186	183	0.496	NUTM1	ENSG00000184507	NON_SYNONYMOUS_CODING	ENST00000438749.3:c.709C>T	ENSP00000407031.3:p.Arg237Cys	2/7	-	-	-	
S14	SNV	chr15	34640808	C	T	369	186	183	0.496	NUTM1	ENSG00000184507	NON_SYNONYMOUS_CODING	ENST00000333756.4:c.655C>T	ENSP00000329448.4:p.Arg219Cys	2/7	YES	-	-	
S18	SNV	chr10	88988322	C	A	96	63	33	0.344	NUTM2A	ENSG00000184923	NON_SYNONYMOUS_CODING	ENST00000381689.4:c.685C>A	ENSP00000371107.3:p.Leu229Ile	2/7	-	0.0004	rs571729371	
S18	SNV	chr10	88988322	C	A	96	63	33	0.344	NUTM2A	ENSG00000184923	NON_SYNONYMOUS_CODING	ENST00000381707.2:c.685C>A	ENSP00000371126.1:p.Leu229Ile	2/7	YES	0.0004	rs571729371	
S27	SNV	chr10	88992480	C	A	33	21	12	0.364	NUTM2A	ENSG00000184923	NON_SYNONYMOUS_CODING	ENST00000381707.2:c.1472C>A	ENSP00000371126.1:p.Thr491Asn	5/7	YES	-	-	
S27	SNV	chr10	88992480	C	A	33	21	12	0.364	NUTM2A	ENSG00000184923	NON_SYNONYMOUS_CODING	ENST00000381689.4:c.1472C>A	ENSP00000371107.3:p.Thr491Asn	5/7	-	-	-	
S25	INDEL	chr9	95179129	G	-A	83	51	32	0.385	OMD	ENSG00000127083	FRAME_SHIFT	ENST00000375550.4:c.712delC	ENSP00000364700.4:p.Leu238CysfsTer3	2/3	YES	-	-	
S48	SNV	chr22	31723284	A	G	523	342	181	0.346	PATZ1	ENSG00000100105	NON_SYNONYMOUS_CODING	ENST00000351933.4:c.1519T>C	ENSP00000337520.4:p.Cys507Arg	4/4	-	-	-	
S48	SNV	chr22	31723284	A	G	523	342	181	0.346	PATZ1	ENSG00000100105	NON_SYNONYMOUS_CODING	ENST00000266269.5:c.1657T>C	ENSP00000266269.5:p.Cys553Arg	5/5	YES	-	-	
S48	SNV	chr22	31723284	A	G	523	342	181	0.346	PATZ1	ENSG00000100105	NON_SYNONYMOUS_CODING	ENST00000405309.3:c.1592T>C	ENSP00000384173.3:p.Met531Thr	5/5	-	-	-	
63	INDEL	chr9	37034092	T	-TTC	34	27	7	0.206	PAX5	ENSG00000196092	UTR_5_PRIME	ENST00000358127.4:c.-58delA		1/10	YES	-	-	
12	INDEL	chr1	18962801	C	-AAG	309	182	127	0.411	PAX7	ENSG00000009709	CODON_CHANGE_PLUS_CODON_DELETION	ENST00000420770.2:c.523delA	ENSP00000403389.2:p.Lys175ArgfsTer39	4/9	-	-	-	
12	INDEL	chr1	18962801	C	-AAG	309	182	127	0.411	PAX7	ENSG00000009709	CODON_CHANGE_PLUS_CODON_DELETION	ENST00000400661.3:c.517delA	ENSP00000383502.3:p.Lys173ArgfsTer39	4/8	-	-	-	
12	INDEL	chr1	18962801	C	-AAG	309	182	127	0.411	PAX7	ENSG00000009709	CODON_CHANGE_PLUS_CODON_DELETION	ENST00000375375.3:c.523delA	ENSP00000364524.3:p.Lys175ArgfsTer39	4/8	YES	-	-	
S15	SNV	chr1	19018326	C	T	238	119	119	0.5	PAX7	ENSG00000009709	NON_SYNONYMOUS_CODING	ENST00000375375.3:c.665C>T	ENSP00000364524.3:p.Thr222Ile	5/8	YES	-	-	
S15	SNV	chr1	19018326	C	T	238	119	119	0.5	PAX7	ENSG00000009709	NON_SYNONYMOUS_CODING	ENST00000400661.3:c.659C>T	ENSP00000383502.3:p.Thr220Ile	5/8	-	-	-	
S15	SNV	chr1	19018326	C	T	238	119	119	0.5	PAX7	ENSG00000009709	NON_SYNONYMOUS_CODING	ENST00000420770.2:c.665C>T	ENSP00000403389.2:p.Thr222Ile	5/9	-	-	-	
47	SNV	chr3	52661307	C	T	112	50	62	0.554	PBRM1	ENSG00000163939	NON_SYNONYMOUS_CODING	ENST00000423351.1:c.1523G>A	ENSP00000387775.1:p.Gly508Asp	13/26	-	-	-	
47	SNV	chr3	52661307	C	T	112	50	62	0.554	PBRM1	ENSG00000163939	NON_SYNONYMOUS_CODING	ENST00000356770.4:c.1427G>A	ENSP00000349213.4:p.Gly476Asp	12/28	-	-	-	
47	SNV	chr3	52661307	C	T	112	50	62	0.554	PBRM1	ENSG00000163939	NON_SYNONYMOUS_CODING	ENST00000446103.1:c.1355G>A	ENSP00000397662.1:p.Gly452Asp	12/20	-	-	-	
47	INDEL	chr3	52597409	T	-ATC	234	121	113	0.483	PBRM1	ENSG00000163939	CODON_CHANGE_PLUS_CODON_DELETION	ENST00000423351.1:c.3973delA	ENSP00000387775.1:p.Ile1325LeufsTer58	24/28	-	-	-	
47	SNV	chr3	52661307	C	T	112	50	62	0.554	PBRM1	ENSG00000163939	NON_SYNONYMOUS_CODING	ENST00000337303.4:c.1523G>A	ENSP00000338302.4:p.Gly508Asp	13/28	-	-	-	
47	SNV	chr3	52661307	C	T	112	50	62	0.554	PBRM1	ENSG00000163939	NON_SYNONYMOUS_CODING	ENST00000409114.3:c.1523G>A	ENSP00000386643.3:p.Gly508Asp	13/30	-	-	-	
47	INDEL	chr3	52597409	T	-ATC	234	121	113	0.483	PBRM1	ENSG00000163939	CODON_CHANGE_PLUS_CODON_DELETION	ENST00000394830.3:c.3901delA	ENSP00000378307.3:p.Ile1301LeufsTer31	25/30	YES	-	-	
47	SNV	chr3	52661307	C	T	112	50	62	0.554	PBRM1	ENSG00000163939	NON_SYNONYMOUS_CODING	ENST00000296302.7:c.1523G>A	ENSP00000296302.7:p.Gly508Asp	13/30	-	-	-	
47	SNV	chr3	52661307	C	T	112	50	62	0.554	PBRM1	ENSG00000163939	NON_SYNONYMOUS_CODING	ENST00000409057.1:c.1523G>A	ENSP00000386593.1:p.Gly508Asp	13/29	-	-	-	
47	SNV	chr3	52661307	C	T	112	50	62	0.554	PBRM1	ENSG00000163939	NON_SYNONYMOUS_CODING	ENST00000410007.1:c.1523G>A	ENSP00000386529.1:p.Gly508Asp	13/29	-	-	-	
47	INDEL	chr3	52597409	T	-ATC	234	121	113	0.483	PBRM1	ENSG00000163939	CODON_CHANGE_PLUS_CODON_DELETION	ENST00000337303.4:c.3976delA	ENSP00000338302.4:p.Ile1326LeufsTer58	24/28	-	-	-	
47	INDEL	chr3	52597409	T	-ATC	234	121	113	0.483	PBRM1	ENSG00000163939	CODON_CHANGE_PLUS_CODON_DELETION	ENST00000409057.1:c.3976delA	ENSP00000386593.1:p.Ile1326LeufsTer58	24/29	-	-	-	

47	INDEL	chr3	52597409	T	-ATC	234	121	113	0.483	PBRM1	ENSG00000163939	CODON_CHANGE_PLUS_CODON_DELETION	ENST00000409767.1:c.4021delA	ENSP00000386601.1:p.Ile1341LeufsTer58	25/29	-	-	-
47	SNV	chr3	52661307	C	T	112	50	62	0.554	PBRM1	ENSG00000163939	NON_SYNONYMOUS_CODING	ENST00000394830.3:c.1523G>A	ENSP00000378307.3:p.Gly508Asp	14/30	YES	-	-
47	INDEL	chr3	52597409	T	-ATC	234	121	113	0.483	PBRM1	ENSG00000163939	CODON_CHANGE_PLUS_CODON_DELETION	ENST00000356770.4:c.3880delA	ENSP00000349213.4:p.Ile1294LeufsTer58	23/28	-	-	-
47	INDEL	chr3	52597409	T	-ATC	234	121	113	0.483	PBRM1	ENSG00000163939	CODON_CHANGE_PLUS_CODON_DELETION	ENST00000409114.3:c.4021delA	ENSP00000386643.3:p.Ile1341LeufsTer58	25/30	-	-	-
47	INDEL	chr3	52597409	T	-ATC	234	121	113	0.483	PBRM1	ENSG00000163939	CODON_CHANGE_PLUS_CODON_DELETION	ENST00000410007.1:c.3901delA	ENSP00000386529.1:p.Ile1301LeufsTer58	24/29	-	-	-
47	INDEL	chr3	52597409	T	-ATC	234	121	113	0.483	PBRM1	ENSG00000163939	CODON_CHANGE_PLUS_CODON_DELETION	ENST00000296302.7:c.3976delA	ENSP00000296302.7:p.Ile1326LeufsTer58	24/30	-	-	-
47	SNV	chr3	52661307	C	T	112	50	62	0.554	PBRM1	ENSG00000163939	NON_SYNONYMOUS_CODING	ENST00000409767.1:c.1523G>A	ENSP00000386601.1:p.Gly508Asp	13/29	-	-	-
12	SNV	chr11	117078379	G	A	461	342	119	0.258	PCSK7	ENSG00000160613	UTR_3_PRIME	ENST00000540028.1:c.*68C>T		7/10	-	-	-
30	SNV	chr1	144857635	G	A	595	450	145	0.244	PDE4DIP	ENSG00000178104	NON_SYNONYMOUS_CODING	ENST00000313382.9:c.6101C>T	ENSP00000327209.9:p.Thr2034Met	41/46	-	0.0002	rs587627879
30	SNV	chr1	144857635	G	A	595	450	145	0.244	PDE4DIP	ENSG00000178104	NON_SYNONYMOUS_CODING	ENST00000530740.1:c.6674C>T	ENSP00000435654.1:p.Thr2225Met	41/46	-	0.0002	rs587627879
30	SNV	chr1	144857635	G	A	595	450	145	0.244	PDE4DIP	ENSG00000178104	NON_SYNONYMOUS_CODING	ENST00000369356.4:c.6419C>T	ENSP00000358363.4:p.Thr2140Met	39/44	YES	0.0002	rs587627879
30	SNV	chr1	144857635	G	A	595	450	145	0.244	PDE4DIP	ENSG00000178104	NON_SYNONYMOUS_CODING	ENST00000369359.4:c.6827C>T	ENSP00000358366.4:p.Thr2276Met	42/47	-	0.0002	rs587627879
30	SNV	chr1	144857635	G	A	595	450	145	0.244	PDE4DIP	ENSG00000178104	NON_SYNONYMOUS_CODING	ENST00000369354.3:c.6419C>T	ENSP00000358360.3:p.Thr2140Met	39/44	-	0.0002	rs587627879
36	SNV	chr1	144879227	G	A	1018	779	239	0.235	PDE4DIP	ENSG00000178104	NON_SYNONYMOUS_CODING	ENST00000369354.3:c.4223C>T	ENSP00000358360.3:p.Ser1408Leu	27/44	-	-	-
36	SNV	chr1	144879227	G	A	1018	779	239	0.235	PDE4DIP	ENSG00000178104	NON_SYNONYMOUS_CODING	ENST00000313382.9:c.4091C>T	ENSP00000327209.9:p.Ser1364Leu	30/46	-	-	-
36	SNV	chr1	144879227	G	A	1018	779	239	0.235	PDE4DIP	ENSG00000178104	NON_SYNONYMOUS_CODING	ENST00000369356.4:c.4223C>T	ENSP00000358363.4:p.Ser1408Leu	27/44	YES	-	-
36	SNV	chr1	144879227	G	A	1018	779	239	0.235	PDE4DIP	ENSG00000178104	NON_SYNONYMOUS_CODING	ENST00000369359.4:c.4631C>T	ENSP00000358366.4:p.Ser1544Leu	30/47	-	-	-
36	SNV	chr1	144879227	G	A	1018	779	239	0.235	PDE4DIP	ENSG00000178104	NON_SYNONYMOUS_CODING	ENST00000530740.1:c.4631C>T	ENSP00000435654.1:p.Ser1544Leu	30/46	-	-	-
48	SNV	chr1	144855853	G	A	452	322	130	0.288	PDE4DIP	ENSG00000178104	NON_SYNONYMOUS_CODING	ENST00000369356.4:c.6700C>T	ENSP00000358363.4:p.Arg2234Trp	41/44	YES	-	rs142861673
48	SNV	chr1	144855853	G	A	452	322	130	0.288	PDE4DIP	ENSG00000178104	NON_SYNONYMOUS_CODING	ENST00000369354.3:c.6700C>T	ENSP00000358360.3:p.Arg2234Trp	41/44	-	-	rs142861673
48	SNV	chr1	144855853	G	A	452	322	130	0.288	PDE4DIP	ENSG00000178104	NON_SYNONYMOUS_CODING	ENST00000313382.9:c.6382C>T	ENSP00000327209.9:p.Arg2128Trp	43/46	-	-	rs142861673
48	SNV	chr1	144855853	G	A	452	322	130	0.288	PDE4DIP	ENSG00000178104	NON_SYNONYMOUS_CODING	ENST00000369359.4:c.7108C>T	ENSP00000358366.4:p.Arg2370Trp	44/47	-	-	rs142861673
48	SNV	chr1	144855853	G	A	452	322	130	0.288	PDE4DIP	ENSG00000178104	NON_SYNONYMOUS_CODING	ENST00000530740.1:c.6955C>T	ENSP00000435654.1:p.Arg2319Trp	43/46	-	-	rs142861673
S34	SNV	chr1	144877123	T	C	970	731	239	0.246	PDE4DIP	ENSG00000178104	NON_SYNONYMOUS_CODING	ENST00000369359.4:c.4972A>G	ENSP00000358366.4:p.Ile1658Val	31/47	-	-	-
S34	SNV	chr1	144877123	T	C	970	731	239	0.246	PDE4DIP	ENSG00000178104	NON_SYNONYMOUS_CODING	ENST00000313382.9:c.4432A>G	ENSP00000327209.9:p.Ile1478Val	31/46	-	-	-
S34	SNV	chr1	144877123	T	C	970	731	239	0.246	PDE4DIP	ENSG00000178104	NON_SYNONYMOUS_CODING	ENST00000369354.3:c.4564A>G	ENSP00000358360.3:p.Ile1522Val	28/44	-	-	-
S34	SNV	chr1	144877123	T	C	970	731	239	0.246	PDE4DIP	ENSG00000178104	NON_SYNONYMOUS_CODING	ENST00000530740.1:c.4972A>G	ENSP00000435654.1:p.Ile1658Val	31/46	-	-	-
S34	SNV	chr1	144877123	T	C	970	731	239	0.246	PDE4DIP	ENSG00000178104	NON_SYNONYMOUS_CODING	ENST00000369356.4:c.4564A>G	ENSP00000358363.4:p.Ile1522Val	28/44	YES	-	-
S44	SNV	chr1	145014096	G	A	1718	1359	359	0.209	PDE4DIP	ENSG00000178104	NON_SYNONYMOUS_CODING	ENST00000493130.2:c.446C>T	ENSP00000432251.1:p.Ala149Val	4/4	-	-	-
S63	SNV	chr1	144909940	C	T	997	767	230	0.23	PDE4DIP	ENSG00000178104	NON_SYNONYMOUS_CODING	ENST00000313382.9:c.2446G>A	ENSP00000327209.9:p.Gly816Arg	20/46	-	-	-
S63	SNV	chr1	144906444	G	C	670	327	343	0.512	PDE4DIP	ENSG00000178104	NON_SYNONYMOUS_CODING	ENST00000313431.9:c.2902C>G	ENSP00000316434.9:p.Gln968Glu	14/19	-	-	-
S63	SNV	chr1	144918931	G	A	1529	1110	419	0.274	PDE4DIP	ENSG00000178104	NON_SYNONYMOUS_CODING	ENST00000369354.3:c.1255C>T	ENSP00000358360.3:p.Arg419Cys	10/44	-	-	rs371722785,COSM4021668,COSM4021669,COSM4021670,COSM4021671,COSM4021667
S63	SNV	chr1	144918931	G	A	1529	1110	419	0.274	PDE4DIP	ENSG00000178104	NON_SYNONYMOUS_CODING	ENST00000530740.1:c.1666C>T	ENSP00000435654.1:p.Arg556Cys	13/46	-	-	rs371722785,COSM4021668,COSM4021669,COSM4021670,COSM4021671,COSM4021667
S63	SNV	chr1	144909940	C	T	997	767	230	0.23	PDE4DIP	ENSG00000178104	NON_SYNONYMOUS_CODING	ENST00000369354.3:c.2248G>A	ENSP00000358360.3:p.Gly750Arg	17/44	-	-	-
S63	SNV	chr1	144909940	C	T	997	767	230	0.23	PDE4DIP	ENSG00000178104	NON_SYNONYMOUS_CODING	ENST00000369349.3:c.2248G>A	ENSP00000358355.3:p.Gly750Arg	17/23	-	-	-
S63	SNV	chr1	144906444	G	C	670	327	343	0.512	PDE4DIP	ENSG00000178104	NON_SYNONYMOUS_CODING	ENST00000369349.3:c.2413C>G	ENSP00000358355.3:p.Gln805Glu	18/23	-	-	-
S63	SNV	chr1	144906444	G	C	670	327	343	0.512	PDE4DIP	ENSG00000178104	NON_SYNONYMOUS_CODING	ENST00000529945.1:c.2902C>G	ENSP00000433392.1:p.Gln968Glu	14/17	-	-	-
S63	SNV	chr1	144909940	C	T	997	767	230	0.23	PDE4DIP	ENSG00000178104	NON_SYNONYMOUS_CODING	ENST00000530740.1:c.2659G>A	ENSP00000435654.1:p.Gly887Arg	20/46	-	-	-
S63	SNV	chr1	144909940	C	T	997	767	230	0.23	PDE4DIP	ENSG00000178104	NON_SYNONYMOUS_CODING	ENST00000529945.1:c.2737G>A	ENSP00000433392.1:p.Gly913Arg	13/17	-	-	-
S63	SNV	chr1	144906444	G	C	670	327	343	0.512	PDE4DIP	ENSG00000178104	NON_SYNONYMOUS_CODING	ENST00000369359.4:c.2824C>G	ENSP00000358366.4:p.Gln942Glu	21/47	-	-	-
S63	SNV	chr1	144909940	C	T	997	767	230	0.23	PDE4DIP	ENSG00000178104	NON_SYNONYMOUS_CODING	ENST00000479408.2:c.1609G>A	ENSP00000436791.1:p.Gly537Arg	13/17	-	-	-
S63	SNV	chr1	144918931	G	A	1529	1110	419	0.274	PDE4DIP	ENSG00000178104	NON_SYNONYMOUS_CODING	ENST00000313382.9:c.1453C>T	ENSP00000327209.9:p.Arg485Cys	13/46	-	-	rs371722785,COSM4021668,COSM4021669,COSM4021670,COSM4021671,COSM4021667
S63	SNV	chr1	144909940	C	T	997	767	230	0.23	PDE4DIP	ENSG00000178104	NON_SYNONYMOUS_CODING	ENST00000369356.4:c.2248G>A	ENSP00000358363.4:p.Gly750Arg	17/44	YES	-	-
S63	SNV	chr1	144906444	G	C	670	327	343	0.512	PDE4DIP	ENSG00000178104	NON_SYNONYMOUS_CODING	ENST00000369351.3:c.2413C>G	ENSP00000358357.3:p.Gln805Glu	18/22	-	-	-
S63	SNV	chr1	144918931	G	A	1529	1110	419	0.274	PDE4DIP	ENSG00000178104	NON_SYNONYMOUS_CODING	ENST00000369356.4:c.1255C>T	ENSP00000358363.4:p.Arg419Cys	10/44	YES	-	rs371722785,COSM4021668,COSM4021669,COSM4021670,COSM4021671,COSM4021667
S63	SNV	chr1	144906444	G	C	670	327	343	0.512	PDE4DIP	ENSG00000178104	NON_SYNONYMOUS_CODING	ENST00000479408.2:c.1774C>G	ENSP00000436791.1:p.Gln592Glu	14/17	-	-	-
S63	SNV	chr1	144918931	G	A	1529	1110	419	0.274	PDE4DIP	ENSG00000178104	NON_SYNONYMOUS_CODING	ENST00000529945.1:c.1744C>T	ENSP00000433392.1:p.Arg582Cys	6/17	-	-	rs371722785,COSM4021668,COSM4021669,COSM4021670,COSM4021671,COSM4021667
S63	SNV	chr1	144906444	G	C	670	327	343	0.512	PDE4DIP	ENSG00000178104	NON_SYNONYMOUS_CODING	ENST00000369354.3:c.2413C>G	ENSP00000358360.3:p.Gln805Glu	18/44	-	-	-
S63	SNV	chr1	144906444	G	C	670	327	343	0.512	PDE4DIP	ENSG00000178104	NON_SYNONYMOUS_CODING	ENST00000530740.1:c.2824C>G	ENSP00000435654.1:p.Gln942Glu	21/46	-	-	-
S63	SNV	chr1	144909940	C	T	997	767	230	0.23	PDE4DIP	ENSG00000178104	NON_SYNONYMOUS_CODING	ENST00000313431.9:c.2737G>A	ENSP00000316434.9:p.Gly913Arg	13/19	-	-	-
S63	SNV	chr1	144918931	G	A	1529	1110	419	0.274	PDE4DIP	ENSG00000178104	NON_SYNONYMOUS_CODING	ENST00000369359.4:c.1666C>T	ENSP00000358366.4:p.Arg556Cys	13/47	-	-	rs371722785,COSM4021668,COSM4021669,COSM4021670,COSM4021671,COSM4021667
S63	SNV	chr1	144918931	G	A	1529	1110	419	0.274	PDE4DIP	ENSG00000178104	NON_SYNONYMOUS_CODING	ENST00000369351.3:c.1255C>T	ENSP00000358357.3:p.Arg419Cys	10/22	-	-	rs371722785,COSM4021668,COSM4021669,COSM4021670,COSM4021671,COSM4021667
S63	SNV	chr1	144918931	G	A	1529	1110	419	0.274	PDE4DIP	ENSG00000178104	NON_SYNONYMOUS_CODING	ENST00000369349.3:c.1255C>T	ENSP00000358355.3:p.Arg419Cys	10/23	-	-	rs371722785,COSM4021668,COSM4021669,COSM4021670,COSM4021671,COSM4021667
S63	SNV	chr1	144906444	G	C	670	327	343	0.512	PDE4DIP	ENSG00000178104	NON_SYNONYMOUS_CODING	ENST00000313382.9:c.2611C>G	ENSP00000327209.9:p.Gln871Glu	21/46	-	-	-
S63	SNV	chr1	144909940	C	T	997	767	230	0.23	PDE4DIP	ENSG00000178104	NON_SYNONYMOUS_CODING	ENST00000369351.3:c.2248G>A	ENSP00000358357.3:p.Gly750Arg	17/22	-	-	-
S63	SNV	chr1	144918931	G	A	1529	1110	419	0.274	PDE4DIP	ENSG00000178104	NON_SYNONYMOUS_CODING	ENST00000313431.9:c.1744C>T	ENSP00000316434.9:p.Arg582Cys	6/19	-	-	rs371722785,COSM4021668,COSM4021669,COSM4021670,COSM4021671,COSM4021667
S63	SNV	chr1	144906444	G	C	670	327	343	0.512	PDE4DIP	ENSG00000178104	NON_SYNONYMOUS_CODING	ENST00000369356.4:c.2413C>G	ENSP00000358363.4:p.Gln805Glu	18/44	YES	-	-
S63	SNV	chr1	144909940	C	T	997	767	230	0.23	PDE4DIP	ENSG00000178104	NON_SYNONYMOUS_CODING	ENST00000369359.4:c.2659G>A	ENSP00000358366.4:p.Gly887Arg	20/47	-	-	-
S63	SNV	chr1	144918931	G	A	1529	1110	419	0.274	PDE4DIP	ENSG00000178104	NON_SYNONYMOUS_CODING	ENST00000479408.2:c.616C>T	ENSP00000436791.1:p.Arg206Cys	6/17	-	-	rs371722785,COSM4021668,COSM4021669,COSM4021670,COSM4021671,COSM4021667
S34	SNV	chr5	149511607	T	C	352	166	186	0.528	PDGFRB	ENSG00000113721	NON_SYNONYMOUS_CODING	ENST00000261799.4:c.1178A>G	ENSP00000261799.4:p.His393Arg	8/23	YES	-	COSM590942
35	SNV	chr17	8050686	G	A	296	171	125	0.422	PER1	ENSG00000179094	NON_SYNONYMOUS_CODING	ENST00000354903.5:c.1463C>T	ENSP00000346979.5:p.Pro488Leu	13/18	-	-	-
35	SNV	chr17	8050686	G	A	296	171	125	0.422	PER1	ENSG00000179094	NON_SYNONYMOUS_CODING	ENST00000317276.4:c.1511C>T	ENSP00000314420.4:p.Pro504Leu	13/23	YES	-	-
42	SNV	chr17	8059644	C	G	407	213	194	0.477	PER1	ENSG00000179094	NON_SYNONYMOUS_CODING	ENST00000354903.5:c.29G>C	ENSP00000346979.5:p.Ser10Thr	1/18	-	0.0004	rs563360327
S43	SNV	chr17	8051107	T	C	210	20	190	0.905	PER1	ENSG00000179094	NON_SYNONYMOUS_CODING	ENST00000354903.5:c.1225A>G	ENSP00000346979.5:p.Ile409Val	11/18	-	-	-
S43	SNV	chr17	8051107	T	C	210	20	190	0.905	PER1	ENSG00000179094	NON_SYNONYMOUS_CODING	ENST00000581082.1:c.1213A>G	ENSP00000462064.1:p.Ile405Val	10/22	-	-	-
S43	SNV	chr17	8051107	T	C	210	20	190	0.905	PER1	ENSG00000179094	NON_SYNONYMOUS_CODING	ENST00000317276.4:c.1273A>G	ENSP00000314420.4:p.Ile425Val	11/23	YES	-	-
S57	SNV	chr17	8047949	T	C	527	266	261	0.495	PER1	ENSG00000179094	NON_SYNONYMOUS_CODING	ENST00000354903.5:c.2533A>G	ENSP00000346979.5:p.Thr845Ala	18/18	-	-	-
S34	SNV	chrX	133549136	C	T	209	110	99	0.474	PHF6	ENSG00000156531	STOP_GAINED	ENST00000370803.3:c.820C>T	ENSP00000359839.3:p.Arg274Ter	8/11	-	-	COSM144567,COSM1134629
S34	SNV	chrX	133549136	C	T	209	110	99	0.474	PHF6	ENSG00000156531	STOP_GAINED	ENST00000416404.2:c.718C>T	ENSP00000394480.2:p.Arg240Ter	7/10	-	-	COSM144567,COSM1134629
S34	SNV	chrX	133549136	C	T	209	110	99	0.474	PHF6	ENSG00000156531	STOP_GAINED	ENST00000332070.3:c.820C>T	ENSP00000329097.3:p.Arg274Ter	8/10	YES	-	COSM144567,COSM1134629
S34	SNV	chrX	133549136	C	T	209	110	99	0.474	PHF6	ENSG00000156531	STOP_GAINED	ENST00000394292.1:c.823C>T	ENSP00000377831.1:p.Arg275Ter	8/11	-	-	COSM144567,COSM1134629
S34	SNV	chrX	133549136	C	T	209	110	99	0.474	PHF6	ENSG00000156531	STOP_GAINED	ENST00000370799.1:c.823C>T	ENSP00000359835.1:p.Arg275Ter	8/9	-	-	COSM144567,COSM1134629
S34	SNV	chrX	133549136	C	T	209	110	99	0.474	PHF6	ENSG00000156531	STOP_GAINED	ENST00000370800.4:c.823C>T	ENSP00000359836.4:p.Arg275Ter	8/8	-	-	COSM144567,COSM1134629
S35	SNV	chrX	133547940	C	T	325	19	306	0.942	PHF6	ENSG00000156531	STOP_GAINED	ENST00000394292.1:c.676C>T	ENSP00000377831.1:p.Arg226Ter	7/11	-	-	COSM255184
S35	SNV	chrX	133547940	C	T	325	19	306	0.942	PHF6	ENSG00000156531	STOP_GAINED	ENST00000416404.2:c.571C>T	ENSP00000394480.2:p.Arg191Ter	6/10	-	-	COSM255184
S35	SNV	chrX	133547940	C	T	325	19	306	0.942	PHF6	ENSG00000156531	STOP_GAINED	ENST00000370800.4:c.676C>T	ENSP00000359836.4:p.Arg226Ter	7/8	-	-	COSM255184
S35	SNV	chrX	133547940	C	T	325	19	306	0.942	PHF6	ENSG00000156531	STOP_GAINED	ENST00000332070.3:c.673C>T	ENSP00000329097.3:p.Arg225Ter	7/10	YES	-	COSM255184
S35	SNV	chrX	133547940	C	T	325	19	306	0.942	PHF6	ENSG00000156531	STOP_GAINED	ENST00000370803.3:c.673C>T	ENSP00000359839.3:p.Arg225Ter	7/11	-	-	COSM255184
S35	SNV	chrX	133547940	C	T	325	19	306	0.942	PHF6	ENSG00000156531	STOP_GAINED	ENST00000370799.1:c.676C>T	ENSP00000359835.1:p.Arg226Ter	7/9	-	-	COSM255184
S36	INDEL	chrX	133547926	A	+G	881	482	399	0.453	PHF6	ENSG00000156531	FRAME_SHIFT	-	-	6/10	-	-	-
S36	INDEL	chrX	133547926	A	+G	881	482	399	0.453	PHF6	ENSG00000156531	FRAME_SHIFT	-	-	7/11	-	-	-
S36	INDEL	chrX	133547926	A	+G	881	482	399	0.453	PHF6	ENSG00000156531	FRAME_SHIFT	-	-	7/8	-	-	-
S36	INDEL	chrX	133547926	A	+G	881	482	399	0.453	PHF6	ENSG00000156531	FRAME_SHIFT	-	-	7/10	YES	-	-
S36	INDEL	chrX	133547926	A	+G	881	482	399	0.453	PHF6	ENSG00000156531	FRAME_SHIFT	-	-	7/9	-	-	-
S36	INDEL	chrX	133547926	A	+G	881	482	399	0.453	PHF6	ENSG00000156531	FRAME_SHIFT	-	-	7/11	-	-	-
S54	SNV	chrX	133551263	C	T	673	360	313	0.465	PHF6	ENSG00000156531	NON_SYNONYMOUS_CODING	ENST00000416404.2:c.797C>T	ENSP00000394480.2:p.Thr266Ile	8/10	-	-	-
S54	SNV	chrX	133551263	C	T	673	360	313	0.465	PHF6	ENSG00000156531	NON_SYNONYMOUS_CODING	ENST00000394292.1:c.902C>T	ENSP00000377831.1:p.Thr301Ile	9/11	-	-	-
S54	SNV	chrX	133551263	C	T	673	360	313	0.465	PHF6	ENSG00000156531	NON_SYNONYMOUS_CODING	ENST00000370803.3:c.899C>T	ENSP00000359839.3:p.Thr300Ile	9/11	-	-	-
S54	SNV	chrX	133551263	C	T	673	360	313	0.465	PHF6	ENSG00000156531	NON_SYNONYMOUS_CODING	ENST00000370799.1:c.902C>T	ENSP00000359835.1:p.Thr301Ile	9/9	-	-	-
S54	SNV	chrX	133551263	C	T	673	360	313	0.465	PHF6	ENSG00000156531	NON_SYNONYMOUS_CODING	ENST00000332070.3:c.899C>T	ENSP00000329097.3:p.Thr300Ile	9/10	YES	-	-
S37	INDEL	chr4	41749672	C	-GA	459	265	194	0.423	PHOX2B	ENSG00000109132	FRAME_SHIFT	ENST00000519424.1:c.533delG	ENSP00000426733.1:p.Arg19AlafsTer55	1/3	-	-	-
46	SNV	chr11	85670067	A	C	62	40	22	0.355	PICALM	ENSG00000073921	UTR_3_PRIME	ENST00000528398.1:c.*22T>G	-	-	19/19	-	-
46	SNV	chr11	85670067	A	C	62	40	22	0.355	PICALM	ENSG00000073921	UTR_3_PRIME	ENST00000356360.5:c.*22T>G	-	-	19/19	-	-
46	SNV	chr11	85670067	A	C	62	40	22	0.355	PICALM	ENSG00000073921	UTR_3_PRIME	ENST00000393346.3:c.*22T>G	-	-	20/20	YES	-
46	SNV	chr11	85670067	A	C	62	40	22	0.355	PICALM	ENSG00000073921	UTR_3_PRIME	ENST00000532317.1:c.*22T>G	-	-	20/20	-	-
46	SNV	chr11	85670067	A	C	62	40	22	0.355	PICALM	ENSG00000073921	UTR_3_PRIME	ENST00000526033.1:c.*22T>G	-	-	20/20	-	-
32	SNV	chr1	204436089	C	T	50	28	22	0.44	PIK3C2B	ENSG00000133056	NON_SYNONYMOUS_CODING	ENST00000367184.2:c.77G>A	ENSP00000356152.2:p.Arg26Gln	1/3	-	-	rs547113789
37	SNV	chr1	204435964	G	A	59	28	31	0.525	PIK3C2B	ENSG00000133056	NON_SYNONYMOUS_CODING	ENST00000367184.2:c.202C>T	ENSP00000356152.2:p.Leu68Phe	1/3	-	0.0008	rs372874064
55	SNV	chr1	204409407	T	A	405	227	178	0.438	PIK3C2B	ENSG00000133056	NON_SYNONYMOUS_CODING	ENST00000367187.3:c.3292A>T	ENSP00000356155.3:p.Ile1098Phe	23/34	YES	-	-

55	SNV	chr1	204409407	T	A	405	227	178	0.438	PIK3C2B	ENSG00000133056	NON_SYNONYMOUS_CODING	ENST00000424712.2:c.3208A>T	ENSP00000400561.2:p.Ile1070Phe	24/35	-	-	-
63	SNV	chr1	204436162	G	A	159	89	70	0.44	PIK3C2B	ENSG00000133056	NON_SYNONYMOUS_CODING	ENST00000367184.2:c.4C>T	ENSP00000356152.2:p.Arg2Trp	1/3	-	-	-
S03	SNV	chr1	204433674	C	G	520	273	247	0.473	PIK3C2B	ENSG00000133056	NON_SYNONYMOUS_CODING	ENST00000367184.2:c.427G>C	ENSP00000356152.2:p.Val143Leu	3/3	-	-	-
S03	SNV	chr1	204433674	C	G	520	273	247	0.473	PIK3C2B	ENSG00000133056	NON_SYNONYMOUS_CODING	ENST00000424712.2:c.1093G>C	ENSP00000400561.2:p.Val365Leu	5/35	-	-	-
S03	SNV	chr1	204433674	C	G	520	273	247	0.473	PIK3C2B	ENSG00000133056	NON_SYNONYMOUS_CODING	ENST00000367187.3:c.1093G>C	ENSP00000356155.3:p.Val365Leu	5/34	YES	-	-
S14	SNV	chr1	204435981	T	G	88	44	44	0.5	PIK3C2B	ENSG00000133056	NON_SYNONYMOUS_CODING	ENST00000367184.2:c.185A>C	ENSP00000356152.2:p.Asp62Ala	1/3	-	0.0002	rs375101836
30	SNV	chr3	178952037	C	T	225	126	99	0.44	PIK3CA	ENSG00000121879	NON_SYNONYMOUS_CODING	ENST00000263967.3:c.3092C>T	ENSP00000263967.3:p.Thr1031Ile	21/21	YES	-	COSM28543
32	SNV	chr15	74287140	C	G	94	45	49	0.521	PML	ENSG00000140464	UTR_5_PRIME	ENST00000435786.2:c.-14C>G	-	1/7	-	-	-
32	SNV	chr15	74287140	C	G	94	45	49	0.521	PML	ENSG00000140464	UTR_5_PRIME	ENST00000436891.3:c.-14C>G	-	1/8	-	-	-
32	SNV	chr15	74287140	C	G	94	45	49	0.521	PML	ENSG00000140464	UTR_5_PRIME	ENST00000359928.4:c.-14C>G	-	1/6	-	-	-
32	SNV	chr15	74287140	C	G	94	45	49	0.521	PML	ENSG00000140464	UTR_5_PRIME	ENST00000395132.2:c.-14C>G	-	1/5	-	-	-
32	SNV	chr15	74287140	C	G	94	45	49	0.521	PML	ENSG00000140464	UTR_5_PRIME	ENST00000395135.3:c.-14C>G	-	1/8	-	-	-
32	SNV	chr15	74287140	C	G	94	45	49	0.521	PML	ENSG00000140464	UTR_5_PRIME	ENST00000268058.3:c.-14C>G	-	1/9	YES	-	-
32	SNV	chr15	74287140	C	G	94	45	49	0.521	PML	ENSG00000140464	UTR_5_PRIME	ENST00000268059.6:c.-14C>G	-	1/8	-	-	-
32	SNV	chr15	74287140	C	G	94	45	49	0.521	PML	ENSG00000140464	UTR_5_PRIME	ENST00000354026.6:c.-14C>G	-	1/7	-	-	-
24	SNV	chr2	190660540	G	A	244	148	96	0.393	PMS1	ENSG00000064933	NON_SYNONYMOUS_CODING	ENST00000447232.2:c.178G>A	ENSP00000401064.2:p.Gly60Ser	3/12	-	-	COSM5728 86
24	SNV	chr2	190660540	G	A	244	148	96	0.393	PMS1	ENSG00000064933	NON_SYNONYMOUS_CODING	ENST00000441310.2:c.178G>A	ENSP00000406490.2:p.Gly60Ser	3/13	YES	-	COSM5728 86
24	SNV	chr2	190660540	G	A	244	148	96	0.393	PMS1	ENSG00000064933	NON_SYNONYMOUS_CODING	ENST00000424766.1:c.178G>A	ENSP00000410082.1:p.Gly60Ser	3/5	-	-	COSM5728 86
24	SNV	chr2	190660540	G	A	244	148	96	0.393	PMS1	ENSG00000064933	NON_SYNONYMOUS_CODING	ENST00000374826.4:c.178G>A	ENSP00000363959.4:p.Gly60Ser	3/5	-	-	COSM5728 86
24	SNV	chr2	190660540	G	A	244	148	96	0.393	PMS1	ENSG00000064933	NON_SYNONYMOUS_CODING	ENST00000409823.3:c.178G>A	ENSP00000387125.3:p.Gly60Ser	3/12	-	-	COSM5728 86
24	SNV	chr2	190660540	G	A	244	148	96	0.393	PMS1	ENSG00000064933	UTR_5_PRIME	ENST00000418224.3:c.-248G>A	-	3/12	-	-	COSM5728 86
24	SNV	chr2	190660540	G	A	244	148	96	0.393	PMS1	ENSG00000064933	NON_SYNONYMOUS_CODING	ENST00000409985.1:c.178G>A	ENSP00000386623.1:p.Gly60Ser	3/4	-	-	COSM5728 86
24	SNV	chr2	190660540	G	A	244	148	96	0.393	PMS1	ENSG00000064933	NON_SYNONYMOUS_CODING	ENST00000420421.1:c.178G>A	ENSP00000391136.1:p.Gly60Ser	2/2	-	-	COSM5728 86
S22	SNV	chr2	190670397	C	T	513	253	260	0.507	PMS1	ENSG00000064933	NON_SYNONYMOUS_CODING	ENST00000447232.2:c.335C>T	ENSP00000401064.2:p.Thr112Met	4/12	-	0.0002	rs576285843
S22	SNV	chr2	190670397	C	T	513	253	260	0.507	PMS1	ENSG00000064933	NON_SYNONYMOUS_CODING	ENST00000424766.1:c.335C>T	ENSP00000410082.1:p.Thr112Met	4/5	-	0.0002	rs576285843
S22	SNV	chr2	190670397	C	T	513	253	260	0.507	PMS1	ENSG00000064933	NON_SYNONYMOUS_CODING	ENST00000424307.1:c.152C>T	ENSP00000389938.1:p.Thr51Met	2/8	-	0.0002	rs576285843
S22	SNV	chr2	190670397	C	T	513	253	260	0.507	PMS1	ENSG00000064933	NON_SYNONYMOUS_CODING	ENST00000409985.1:c.335C>T	ENSP00000386623.1:p.Thr112Met	4/4	-	0.0002	rs576285843
S22	SNV	chr2	190670397	C	T	513	253	260	0.507	PMS1	ENSG00000064933	NON_SYNONYMOUS_CODING	ENST00000409823.3:c.335C>T	ENSP00000387125.3:p.Thr112Met	4/12	-	0.0002	rs576285843
S22	SNV	chr2	190670397	C	T	513	253	260	0.507	PMS1	ENSG00000064933	NON_SYNONYMOUS_CODING	ENST00000374826.4:c.335C>T	ENSP00000363959.4:p.Thr112Met	4/5	-	0.0002	rs576285843
S22	SNV	chr2	190670397	C	T	513	253	260	0.507	PMS1	ENSG00000064933	NON_SYNONYMOUS_CODING	ENST00000441310.2:c.335C>T	ENSP00000406490.2:p.Thr112Met	4/13	YES	0.0002	rs576285843
S35	SNV	chr2	190719754	C	T	656	325	331	0.505	PMS1	ENSG00000064933	NON_SYNONYMOUS_CODING	ENST00000418224.3:c.1228C>T	ENSP00000404492.3:p.Arg410Cys	8/12	-	0.0002	rs566975683,COSM3575566
S35	SNV	chr2	190719754	C	T	656	325	331	0.505	PMS1	ENSG00000064933	NON_SYNONYMOUS_CODING	ENST00000447232.2:c.1756C>T	ENSP00000401064.2:p.Arg586Cys	9/12	-	0.0002	rs566975683,COSM3575566
S35	SNV	chr2	190719754	C	T	656	325	331	0.505	PMS1	ENSG00000064933	NON_SYNONYMOUS_CODING	ENST00000424307.1:c.1573C>T	ENSP00000389938.1:p.Arg525Cys	7/8	-	0.0002	rs566975683,COSM3575566
S35	SNV	chr2	190719754	C	T	656	325	331	0.505	PMS1	ENSG00000064933	NON_SYNONYMOUS_CODING	ENST00000432292.3:c.1228C>T	ENSP00000398378.3:p.Arg410Cys	7/11	-	0.0002	rs566975683,COSM3575566
S35	SNV	chr2	190719754	C	T	656	325	331	0.505	PMS1	ENSG00000064933	NON_SYNONYMOUS_CODING	ENST00000409823.3:c.1639C>T	ENSP00000387125.3:p.Arg547Cys	8/12	-	0.0002	rs566975683,COSM3575566
S35	SNV	chr2	190719754	C	T	656	325	331	0.505	PMS1	ENSG00000064933	NON_SYNONYMOUS_CODING	ENST00000441310.2:c.1756C>T	ENSP00000406490.2:p.Arg586Cys	9/13	YES	0.0002	rs566975683,COSM3575566
S35	SNV	chr2	190719754	C	T	656	325	331	0.505	PMS1	ENSG00000064933	NON_SYNONYMOUS_CODING	ENST00000409593.1:c.1111C>T	ENSP00000387169.1:p.Arg371Cys	4/7	-	0.0002	rs566975683,COSM3575566
1	SNV	chr7	72412504	C	A	106	63	43	0.406	POM121	ENSG00000196313	NON_SYNONYMOUS_CODING	ENST00000434423.2:c.1972C>A	ENSP00000405562.2:p.Pro658Thr	11/13	-	-	-
1	SNV	chr7	72412504	C	A	106	63	43	0.406	POM121	ENSG00000196313	NON_SYNONYMOUS_CODING	ENST00000358357.3:c.1177C>A	ENSP00000351124.3:p.Pro393Thr	11/13	-	-	-
1	SNV	chr7	72412504	C	A	106	63	43	0.406	POM121	ENSG00000196313	NON_SYNONYMOUS_CODING	ENST00000257622.4:c.1177C>A	ENSP00000257622.4:p.Pro393Thr	13/15	-	-	-
1	SNV	chr7	72412504	C	A	106	63	43	0.406	POM121	ENSG00000196313	NON_SYNONYMOUS_CODING	ENST00000395270.1:c.1177C>A	ENSP00000378687.1:p.Pro393Thr	14/16	YES	-	-
1	SNV	chr7	72412504	C	A	106	63	43	0.406	POM121	ENSG00000196313	NON_SYNONYMOUS_CODING	ENST00000446813.1:c.1177C>A	ENSP00000393020.1:p.Pro393Thr	13/15	-	-	-
42	SNV	chr7	72412540	A	G	251	168	83	0.331	POM121	ENSG00000196313	NON_SYNONYMOUS_CODING	ENST00000446813.1:c.1213A>G	ENSP00000393020.1:p.Met405Val	13/15	-	-	-
42	SNV	chr7	72412540	A	G	251	168	83	0.331	POM121	ENSG00000196313	NON_SYNONYMOUS_CODING	ENST00000257622.4:c.1213A>G	ENSP00000257622.4:p.Met405Val	13/15	-	-	-
42	SNV	chr7	72412540	A	G	251	168	83	0.331	POM121	ENSG00000196313	NON_SYNONYMOUS_CODING	ENST00000434423.2:c.2008A>G	ENSP00000405562.2:p.Met670Val	11/13	-	-	-
42	SNV	chr7	72412540	A	G	251	168	83	0.331	POM121	ENSG00000196313	NON_SYNONYMOUS_CODING	ENST00000358357.3:c.1213A>G	ENSP00000351124.3:p.Met405Val	11/13	-	-	-
42	SNV	chr7	72412540	A	G	251	168	83	0.331	POM121	ENSG00000196313	NON_SYNONYMOUS_CODING	ENST00000395270.1:c.1213A>G	ENSP00000378687.1:p.Met405Val	14/16	YES	-	-
S15	INDEL	chr7	72395725	C	-GGCGG	32	8	24	0.75	POM121	ENSG00000196313	CODON_CHANGE_PLUS_CODON_DELETION	ENST00000434423.2:c.67delG	ENSP00000405562.2:p.Gly23AlafsTer36	1/13	-	-	-
S29	SNV	chr7	72395869	C	G	130	56	74	0.569	POM121	ENSG00000196313	NON_SYNONYMOUS_CODING	ENST00000434423.2:c.210C>G	ENSP00000405562.2:p.Ser70Arg	1/13	-	-	-
55	SNV	chr1	156737757	C	T	65	26	39	0.6	PRCC	ENSG00000143294	NON_SYNONYMOUS_CODING	ENST00000271526.4:c.194C>T	ENSP00000271526.4:p.Pro65Leu	1/7	YES	0.0004	rs369713102
55	SNV	chr1	156737757	C	T	65	26	39	0.6	PRCC	ENSG00000143294	NON_SYNONYMOUS_CODING	ENST00000353233.3:c.194C>T	ENSP00000339300.3:p.Pro65Leu	1/6	-	0.0004	rs369713102
S57	SNV	chr1	156737838	C	A	289	115	174	0.602	PRCC	ENSG00000143294	NON_SYNONYMOUS_CODING	ENST00000271526.4:c.275C>A	ENSP00000271526.4:p.Pro92Gln	1/7	YES	-	-
S57	SNV	chr1	156737838	C	A	289	115	174	0.602	PRCC	ENSG00000143294	NON_SYNONYMOUS_CODING	ENST00000353233.3:c.275C>A	ENSP00000339300.3:p.Pro92Gln	1/6	-	-	-
63	SNV	chr6	106553012	G	A	446	235	211	0.473	PRDM1	ENSG00000057657	NON_SYNONYMOUS_CODING	ENST00000369089.3:c.575G>A	ENSP00000358098.5:p.Arg192His	3/5	-	-	COSM1262 935
63	SNV	chr6	106553012	G	A	446	235	211	0.473	PRDM1	ENSG00000057657	NON_SYNONYMOUS_CODING	ENST00000369091.2:c.869G>A	ENSP00000358087.2:p.Arg290His	5/7	-	-	COSM1262 935
63	SNV	chr6	106553012	G	A	446	235	211	0.473	PRDM1	ENSG00000057657	NON_SYNONYMOUS_CODING	ENST00000369096.4:c.977G>A	ENSP00000358092.4:p.Arg326His	5/7	YES	-	COSM1262 935
S53	SNV	chr1	3102961	A	C	462	245	217	0.47	PRDM16	ENSG00000142611	NON_SYNONYMOUS_CODING	ENST00000514189.1:c.310A>C	ENSP00000421400.1:p.Met104Leu	2/16	-	-	-
S53	SNV	chr1	3102961	A	C	462	245	217	0.47	PRDM16	ENSG00000142611	NON_SYNONYMOUS_CODING	ENST00000442529.2:c.310A>C	ENSP00000405253.2:p.Met104Leu	2/17	-	-	-
S53	SNV	chr1	3102961	A	C	462	245	217	0.47	PRDM16	ENSG00000142611	NON_SYNONYMOUS_CODING	ENST00000441472.2:c.310A>C	ENSP00000407968.2:p.Met104Leu	2/17	-	-	-
S53	SNV	chr1	3102961	A	C	462	245	217	0.47	PRDM16	ENSG00000142611	NON_SYNONYMOUS_CODING	ENST00000378391.2:c.310A>C	ENSP00000367643.2:p.Met104Leu	2/17	-	-	-
S53	SNV	chr1	3102961	A	C	462	245	217	0.47	PRDM16	ENSG00000142611	NON_SYNONYMOUS_CODING	ENST00000511072.1:c.310A>C	ENSP00000426975.1:p.Met104Leu	2/16	-	-	-
S53	SNV	chr1	3102961	A	C	462	245	217	0.47	PRDM16	ENSG00000142611	NON_SYNONYMOUS_CODING	ENST00000378398.3:c.310A>C	ENSP00000367651.3:p.Met104Leu	2/18	-	-	-
S53	SNV	chr1	3102961	A	C	462	245	217	0.47	PRDM16	ENSG00000142611	NON_SYNONYMOUS_CODING	ENST00000270722.5:c.310A>C	ENSP00000270722.5:p.Met104Leu	2/17	YES	-	-
6	SNV	chr10	72360198	G	C	388	278	110	0.284	PRF1	ENSG00000180644	NON_SYNONYMOUS_CODING	ENST00000441259.1:c.461C>G	ENSP00000398568.1:p.Ala154Gly	2/3	YES	-	-
6	SNV	chr10	72360198	G	C	388	278	110	0.284	PRF1	ENSG00000180644	NON_SYNONYMOUS_CODING	ENST00000373209.2:c.461C>G	ENSP00000362305.1:p.Ala154Gly	2/3	-	-	-
19	SNV	chr9	98209390	G	A	176	104	72	0.409	PTCH1	ENSG00000185920	NON_SYNONYMOUS_CODING	ENST00000430669.2:c.3950C>T	ENSP00000410287.2:p.Pro1317Leu	23/23	-	0.0002	rs571030658
19	SNV	chr9	98209390	G	A	176	104	72	0.409	PTCH1	ENSG00000185920	NON_SYNONYMOUS_CODING	ENST00000437951.1:c.3950C>T	ENSP00000389744.1:p.Pro1317Leu	23/24	-	0.0002	rs571030658
19	SNV	chr9	98209390	G	A	176	104	72	0.409	PTCH1	ENSG00000185920	NON_SYNONYMOUS_CODING	ENST00000375274.2:c.4145C>T	ENSP00000364223.2:p.Pro1382Leu	23/24	-	0.0002	rs571030658
19	SNV	chr9	98209390	G	A	176	104	72	0.409	PTCH1	ENSG00000185920	NON_SYNONYMOUS_CODING	ENST00000421141.1:c.3695C>T	ENSP00000399981.1:p.Pro1232Leu	23/24	-	0.0002	rs571030658
19	SNV	chr9	98209390	G	A	176	104	72	0.409	PTCH1	ENSG00000185920	NON_SYNONYMOUS_CODING	ENST00000331920.6:c.4148C>T	ENSP00000332353.6:p.Pro1383Leu	23/24	YES	0.0002	rs571030658
19	SNV	chr9	98209390	G	A	176	104	72	0.409	PTCH1	ENSG00000185920	NON_SYNONYMOUS_CODING	ENST00000418258.1:c.3695C>T	ENSP00000396135.1:p.Pro1232Leu	23/24	-	0.0002	rs571030658
19	SNV	chr9	98209390	G	A	176	104	72	0.409	PTCH1	ENSG00000185920	NON_SYNONYMOUS_CODING	ENST00000429896.2:c.3695C>T	ENSP00000414823.2:p.Pro1232Leu	23/24	-	0.0002	rs571030658
37	SNV	chr9	98209366	C	T	60	29	31	0.517	PTCH1	ENSG00000185920	NON_SYNONYMOUS_CODING	ENST00000418258.1:c.3719G>A	ENSP00000396135.1:p.Arg1240Gln	23/24	-	-	COSM3746113,COSM3746114,COSM3746116,COSM3746115
37	SNV	chr9	98209366	C	T	60	29	31	0.517	PTCH1	ENSG00000185920	NON_SYNONYMOUS_CODING	ENST00000331920.6:c.4172G>A	ENSP00000332353.6:p.Arg1391Gln	23/24	YES	-	COSM3746113,COSM3746114,COSM3746116,COSM3746115
37	SNV	chr9	98209366	C	T	60	29	31	0.517	PTCH1	ENSG00000185920	NON_SYNONYMOUS_CODING	ENST00000437951.1:c.3974G>A	ENSP00000389744.1:p.Arg1325Gln	23/24	-	-	COSM3746113,COSM3746114,COSM3746116,COSM3746115
37	SNV	chr9	98209366	C	T	60	29	31	0.517	PTCH1	ENSG00000185920	NON_SYNONYMOUS_CODING	ENST00000421141.1:c.3719G>A	ENSP00000399981.1:p.Arg1240Gln	23/24	-	-	COSM3746113,COSM3746114,COSM3746116,COSM3746115
37	SNV	chr9	98209366	C	T	60	29	31	0.517	PTCH1	ENSG00000185920	NON_SYNONYMOUS_CODING	ENST00000429896.2:c.3719G>A	ENSP00000414823.2:p.Arg1240Gln	23/24	-	-	COSM3746113,COSM3746114,COSM3746116,COSM3746115
37	SNV	chr9	98209366	C	T	60	29	31	0.517	PTCH1	ENSG00000185920	NON_SYNONYMOUS_CODING	ENST00000430669.2:c.3974G>A	ENSP00000410287.2:p.Arg1325Gln	23/23	-	-	COSM3746113,COSM3746114,COSM3746116,COSM3746115
37	SNV	chr9	98209366	C	T	60	29	31	0.517	PTCH1	ENSG00000185920	NON_SYNONYMOUS_CODING	ENST00000375274.2:c.4169G>A	ENSP00000364223.2:p.Arg1390Gln	23/24	-	-	COSM3746113,COSM3746114,COSM3746116,COSM3746115
S03	SNV	chr9	98239124	C	T	549	319	230	0.419	PTCH1	ENSG00000185920	NON_SYNONYMOUS_CODING	ENST00000375274.2:c.1516G>A	ENSP00000364223.2:p.Ala506Thr	11/24	-	-	-
S03	SNV	chr9	98239124	C	T	549	319	230	0.419	PTCH1	ENSG00000185920	NON_SYNONYMOUS_CODING	ENST00000430669.2:c.1321G>A	ENSP00000410287.2:p.Ala441Thr	11/23	-	-	-
S03	SNV	chr9	98239124	C	T	549	319	230	0.419	PTCH1	ENSG00000185920	NON_SYNONYMOUS_CODING	ENST00000418258.1:c.1066G>A	ENSP00000396135.1:p.Ala356Thr	11/24	-	-	-
S03	SNV	chr9	98239124	C	T	549	319	230	0.419	PTCH1	ENSG00000185920	NON_SYNONYMOUS_CODING	ENST00000429896.2:c.1066G>A	ENSP00000414823.2:p.Ala356Thr	11/24	-	-	-
S03	SNV	chr9	98239124	C	T	549	319	230	0.419	PTCH1	ENSG00000185920	NON_SYNONYMOUS_CODING	ENST00000437951.1:c.1321G>A	ENSP00000389744.1:p.Ala441Thr	11/24	-	-	-
S03	SNV	chr9	98239124	C	T	549	319	230	0.419	PTCH1	ENSG00000185920	NON_SYNONYMOUS_CODING	ENST00000331920.6:c.1519G>A	ENSP00000332353.6:p.Ala507Thr	11/24	YES	-	-
S03	SNV	chr9	98239124	C	T	549	319	230	0.419	PTCH1	ENSG00000185920	NON_SYNONYMOUS_CODING	ENST00000421141.1:c.1066G>A	ENSP00000399981.1:p.Ala356Thr	11/24	-	-	-
S03	SNV	chr9	98239124	C	T	549	319	230	0.419	PTCH1	ENSG00000185920	NON_SYNONYMOUS_CODING	ENST00000375271.4:c.514G>A	ENSP00000364420.4:p.Ala172Thr	5/8	-	-	-
S29	SNV	chr9	98224206	C	T	369	193	176	0.476	PTCH1	ENSG00000185920	NON_SYNONYMOUS_CODING	ENST00000437951.1:c.2437G>A	ENSP00000389744.1:p.Asp813Asn	16/24	-	-	COSM27115,COSM95096,COSM95097,COSM95098
S29	SNV	chr9	98224206	C	T	369	193	176	0.476	PTCH1	ENSG00000185920	NON_SYNONYMOUS_CODING	ENST00000429896.2:c.2182G>A	ENSP00000414823.2:p.Asp728Asn	16/24	-	-	COSM27115,COSM95096,COSM95097,COSM95098
S29	SNV	chr9	98224206	C	T	369	193	176	0.476	PTCH1	ENSG00000185920	NON_SYNONYMOUS_CODING	ENST00000418258.1:c.2182G>A	ENSP00000396135.1:p.Asp728Asn	16/24	-	-	COSM27115,COSM95096,COSM95097,COSM95098
S29	SNV	chr9	98224206	C	T	369	193	176	0.476	PTCH1	ENSG00000185920	NON_SYNONYMOUS_CODING	ENST00000375274.2:c.2632G>A	ENSP00000364223.2:p.Asp878Asn	16/24	-	-	COSM27115,COSM95096,COSM95097,COSM95098
S29	SNV	chr9	98224206	C	T	369	193	176	0.476	PTCH1	ENSG00000185920	NON_SYNONYMOUS_CODING	ENST00000331920.6:c.2635G>A	ENSP00000332353.6:p.Asp879Asn	16/24	YES	-	COSM27115,COSM95096,COSM95097,COSM95098
S29	SNV	chr9	98224206	C	T	369	193	176	0.476	PTCH1	ENSG00000185920	NON_SYNONYMOUS_CODING	ENST00000430669.2:c.2437G>A	ENSP00000410287.2:p.Asp813Asn	16/23	-	-	COSM27115,COSM95096,COSM95097,COSM95098
S29	SNV	chr9	98224206	C	T	369	193	176	0.476	PTCH1	ENSG00000185920	NON_SYNONYMOUS_CODING	ENST00000421141.1:c.2182G>A	ENSP00000399981.1:p.Asp728Asn	16/24	-	-	COSM27115,COSM95096,COSM95097,COSM95098
46	SNV	chr12	112888199	C	A	157	90	67	0.427	PTPN11	ENSG00000179295	NON_SYNONYMOUS_CODING	ENST00000351677.2:c.215C>A	ENSP00000340944.2:p.Ala72Asp	3/16	YES	-	rs121918454,COSM13035,COSM13015,CM013417
46	SNV	chr12	112888199	C	A	157	90	67	0.427	PTPN11	ENSG00000179295	NON_SYNONYMOUS_CODING	ENST00000392597.1:c.215C>A	ENSP00000376376.1:p.Ala72Asp	3/11	-	-	rs121918454,COSM13035,COSM13015,CM013417
S12	SNV	chr20	40713365	C	T	203	94	109	0.537	PTPRT	ENSG00000196090	NON_SYNONYMOUS_CODING	ENST00000373184.1:c.4123G>A	ENSP00000362280.1:p.Glu1375Lys	29/31	-	-	-
S12	SNV	chr20	40713365	C	T	203	94	109	0.537	PTPRT	ENSG00000196090	NON_SYNONYMOUS_CODING	ENST00000356100.2:c.4120G>A	ENSP00000348408.2:p.Glu1374Lys	29/31	-	-	-
S12	SNV	chr20	40713365	C	T	203	94	109	0.537	PTPRT	ENSG00000196090	NON_SYNONYMOUS_CODING	ENST00000373201.1:c.4063G>A	ENSP00000362297.1:p.Glu1355Lys	28/30	-	-	-

S12	SNV	chr20	40713365	C	T	203	94	109	0.537	PTPRT	ENSG00000196090	NON_SYNONYMOUS_CODING	ENST00000373190.1:c.4090G>A	ENSP00000362286.1:p.Glu1364Lys	29/31	-	-	-
S12	SNV	chr20	40713365	C	T	203	94	109	0.537	PTPRT	ENSG00000196090	NON_SYNONYMOUS_CODING	ENST00000373198.4:c.4150G>A	ENSP00000362294.4:p.Glu1384Lys	30/32	-	-	-
S12	SNV	chr20	40713365	C	T	203	94	109	0.537	PTPRT	ENSG00000196090	NON_SYNONYMOUS_CODING	ENST00000373193.3:c.4102G>A	ENSP00000362289.3:p.Glu1368Lys	29/31	-	-	-
S12	SNV	chr20	40713365	C	T	203	94	109	0.537	PTPRT	ENSG00000196090	NON_SYNONYMOUS_CODING	ENST00000373187.1:c.4093G>A	ENSP00000362283.1:p.Glu1365Lys	29/31	YES	-	-
S25	SNV	chr3	12645699	G	A	97	58	39	0.402	RAF1	ENSG00000132155	NON_SYNONYMOUS_CODING	ENST00000432427.2:c.407C>T	ENSP00000398591.2:p.Ser136Leu	4/14	-	-	-
S25	SNV	chr3	12645699	G	A	97	58	39	0.402	RAF1	ENSG00000132155	NON_SYNONYMOUS_CODING	ENST00000251849.4:c.770C>T	ENSP00000251849.4:p.Ser257Leu	7/17	YES	-	rs80338796,COSM181063,COSM581519,COSM3586964,CM073301
S25	SNV	chr3	12645699	G	A	97	58	39	0.402	RAF1	ENSG00000132155	NON_SYNONYMOUS_CODING	ENST00000442415.2:c.770C>T	ENSP00000401888.2:p.Ser257Leu	7/18	-	-	rs80338796,COSM181063,COSM581519,COSM3586964,CM073301
S25	SNV	chr3	12645699	G	A	97	58	39	0.402	RAF1	ENSG00000132155	NON_SYNONYMOUS_CODING	ENST00000542177.1:c.527C>T	ENSP00000443567.1:p.Ser176Leu	6/16	-	-	rs80338796,COSM181063,COSM581519,COSM3586964,CM073301
S25	SNV	chr3	12645699	G	A	97	58	39	0.402	RAF1	ENSG00000132155	NON_SYNONYMOUS_CODING	ENST00000534997.1:c.125C>T	ENSP00000441186.1:p.Ser42Leu	3/13	-	-	rs80338796,COSM181063,COSM581519,COSM3586964,CM073301
S65	SNV	chr9	135996490	G	A	62	34	28	0.452	RALGDS	ENSG00000160271	NON_SYNONYMOUS_CODING	ENST00000372047.3:c.52C>T	ENSP00000361117.3:p.Pro18Ser	1/18	-	-	-
S65	SNV	chr9	135996490	G	A	62	34	28	0.452	RALGDS	ENSG00000160271	NON_SYNONYMOUS_CODING	ENST00000372050.3:c.52C>T	ENSP00000361120.3:p.Pro18Ser	1/18	YES	-	-
S44	SNV	chr5	170610229	C	T	388	212	176	0.454	RANBP17	ENSG00000204764	NON_SYNONYMOUS_CODING	ENST00000523189.1:c.1924C>T	ENSP00000427975.1:p.His642Tyr	17/28	YES	-	-
55	SNV	chr2	109367845	G	A	479	239	240	0.501	RANBP2	ENSG00000153201	NON_SYNONYMOUS_CODING	ENST00000283195.6:c.1399G>A	ENSP00000283195.6:p.Glu467Lys	10/29	YES	-	-
S25	INDEL	chr2	109383942	T	-A	39	29	10	0.256	RANBP2	ENSG00000153201	FRAME_SHIFT	ENST00000283195.6:c.6948delA	ENSP00000283195.6:p.Leu2316PhefsTer4	20/29	YES	-	-
S25	SNV	chr2	109383921	A	T	33	23	10	0.303	RANBP2	ENSG00000153201	NON_SYNONYMOUS_CODING	ENST00000283195.6:c.6926A>T	ENSP00000283195.6:p.Tyr2309Phe	20/29	YES	-	-
S25	SNV	chr2	109383944	C	T	39	29	10	0.256	RANBP2	ENSG00000153201	NON_SYNONYMOUS_CODING	ENST00000283195.6:c.6949C>T	ENSP00000283195.6:p.Pro2317Ser	20/29	YES	-	-
38	SNV	chr17	38512515	C	T	323	158	165	0.511	RARA	ENSG00000131759	UTR_3_PRIME	ENST00000425707.3:c.*37C>T	-	7/7	-	0.0002	rs543619460
38	SNV	chr17	38512515	C	T	323	158	165	0.511	RARA	ENSG00000131759	UTR_3_PRIME	ENST00000394089.2:c.*37C>T	-	9/9	-	0.0002	rs543619460
38	SNV	chr17	38512515	C	T	323	158	165	0.511	RARA	ENSG00000131759	UTR_3_PRIME	ENST00000254066.5:c.*37C>T	-	9/9	YES	0.0002	rs543619460
38	SNV	chr17	38512515	C	T	323	158	165	0.511	RARA	ENSG00000131759	UTR_3_PRIME	ENST00000394081.3:c.*37C>T	-	8/8	-	0.0002	rs543619460
38	SNV	chr17	38512515	C	T	323	158	165	0.511	RARA	ENSG00000131759	UTR_3_PRIME	ENST00000394086.3:c.*37C>T	-	8/8	-	0.0002	rs543619460
S29	SNV	chr17	38512390	G	T	135	57	78	0.578	RARA	ENSG00000131759	NON_SYNONYMOUS_CODING	ENST00000394089.2:c.1301G>T	ENSP00000377649.2:p.Gly434Val	9/9	-	0.0004	rs553921530
S29	SNV	chr17	38512390	G	T	135	57	78	0.578	RARA	ENSG00000131759	NON_SYNONYMOUS_CODING	ENST00000425707.3:c.1010G>T	ENSP00000389993.3:p.Gly337Val	7/7	-	0.0004	rs553921530
S29	SNV	chr17	38512390	G	T	135	57	78	0.578	RARA	ENSG00000131759	NON_SYNONYMOUS_CODING	ENST00000254066.5:c.1301G>T	ENSP00000254066.5:p.Gly434Val	9/9	YES	0.0004	rs553921530
S29	SNV	chr17	38512390	G	T	135	57	78	0.578	RARA	ENSG00000131759	NON_SYNONYMOUS_CODING	ENST00000394081.3:c.1286G>T	ENSP00000377643.3:p.Gly429Val	8/8	-	0.0004	rs553921530
S29	SNV	chr17	38512390	G	T	135	57	78	0.578	RARA	ENSG00000131759	NON_SYNONYMOUS_CODING	ENST00000394086.3:c.1349G>T	ENSP00000377648.3:p.Gly450Val	8/8	-	0.0004	rs553921530
S64	SNV	chr13	49030384	C	T	845	444	401	0.475	RB1	ENSG00000139687	NON_SYNONYMOUS_CODING	ENST00000267163.4:c.1859C>T	ENSP00000267163.4:p.Thr620Met	19/27	YES	-	rs554834063
35	SNV	chr1	110882106	G	A	287	145	142	0.495	RBM15	ENSG00000162775	NON_SYNONYMOUS_CODING	ENST00000487146.2:c.79G>A	ENSP00000473552.1:p.Ala27Thr	1/2	-	-	-
35	SNV	chr1	110882106	G	A	287	145	142	0.495	RBM15	ENSG00000162775	NON_SYNONYMOUS_CODING	ENST00000369784.3:c.79G>A	ENSP00000358799.3:p.Ala27Thr	1/3	YES	-	-
S10	INDEL	chr1	110882648	G	-TCGGGT	285	183	102	0.358	RBM15	ENSG00000162775	CODON_CHANGE_PLUS_CODON_DELETION	ENST00000369784.3:c.622delT	ENSP00000358799.3:p.Ser208ArgfsTer10	1/3	YES	-	-
S10	INDEL	chr1	110882648	G	-TCGGGT	285	183	102	0.358	RBM15	ENSG00000162775	CODON_CHANGE_PLUS_CODON_DELETION	ENST00000487146.2:c.622delT	ENSP00000473552.1:p.Ser208ArgfsTer10	1/2	-	-	-
S10	INDEL	chr1	110882648	G	-TCGGGT	285	183	102	0.358	RBM15	ENSG00000162775	CODON_CHANGE_PLUS_CODON_DELETION	ENST00000602849.1:c.622delT	ENSP00000473638.1:p.Ser208ArgfsTer10	1/1	-	-	-
24	INDEL	chr8	145738410	T	+GGTGCA	85	66	19	0.224	RECQL4	ENSG00000160957	CODON_INSERTION	-	-	16/22	YES	-	-
24	INDEL	chr8	145738410	T	+GGTGCA	85	66	19	0.224	RECQL4	ENSG00000160957	CODON_INSERTION	-	-	6/8	-	-	-
47	SNV	chr8	145739069	G	T	240	130	110	0.458	RECQL4	ENSG00000160957	NON_SYNONYMOUS_CODING	ENST00000534626.2:c.455C>A	ENSP00000477457.1:p.Arg153Ser	4/8	-	0.0024	rs531970883
47	SNV	chr8	145739069	G	T	240	130	110	0.458	RECQL4	ENSG00000160957	NON_SYNONYMOUS_CODING	ENST00000428558.2:c.2086C>A	ENSP00000475456.1:p.Arg696Ser	13/22	YES	0.0024	rs531970883
6	SNV	chr8	145738998	G	A	394	227	167	0.424	RECQL4	ENSG00000160957	NON_SYNONYMOUS_CODING	ENST00000534626.2:c.526C>T	ENST00000534626.2:c.526C>T(p.%3D)	4/8	-	-	-
6	SNV	chr8	145738998	G	A	394	227	167	0.424	RECQL4	ENSG00000160957	NON_SYNONYMOUS_CODING	ENST00000428558.2:c.2157C>T	ENST00000428558.2:c.2157C>T(p.%3D)	13/22	YES	-	-
S54	SNV	chr8	145738639	C	T	234	115	119	0.509	RECQL4	ENSG00000160957	NON_SYNONYMOUS_CODING	ENST00000428558.2:c.2425G>A	ENSP00000475456.1:p.Gly809Arg	15/22	YES	-	-
S62	SNV	chr8	145738644	C	T	198	88	110	0.556	RECQL4	ENSG00000160957	NON_SYNONYMOUS_CODING	ENST00000428558.2:c.2420G>A	ENSP00000475456.1:p.Arg807His	15/22	YES	0.0002	rs571684398
S46	SNV	chr2	61118914	C	T	650	371	279	0.429	REL	ENSG00000162924	NON_SYNONYMOUS_CODING	ENST00000295025.8:c.107C>T	ENSP00000295025.7:p.Pro36Leu	2/11	YES	-	-
S46	SNV	chr2	61118914	C	T	650	371	279	0.429	REL	ENSG00000162924	NON_SYNONYMOUS_CODING	ENST00000394479.3:c.107C>T	ENSP00000377989.3:p.Pro36Leu	2/10	-	-	-
21	SNV	chr10	43597846	C	A	380	208	172	0.453	RET	ENSG00000165731	NON_SYNONYMOUS_CODING	ENST00000340058.5:c.394C>A	ENSP00000344798.4:p.Leu132Ile	3/19	-	-	-
21	SNV	chr10	43597846	C	A	380	208	172	0.453	RET	ENSG00000165731	NON_SYNONYMOUS_CODING	ENST00000355710.3:c.394C>A	ENSP00000347942.3:p.Leu132Ile	3/20	YES	-	-
38	SNV	chr10	43596063	G	A	272	131	141	0.518	RET	ENSG00000165731	NON_SYNONYMOUS_CODING	ENST00000355710.3:c.230G>A	ENSP00000347942.3:p.Arg77His	2/20	YES	0.0004	rs570176656,COSM294752,COSM538683,COSM1648717
38	SNV	chr10	43596063	G	A	272	131	141	0.518	RET	ENSG00000165731	NON_SYNONYMOUS_CODING	ENST00000340058.5:c.230G>A	ENSP00000344798.4:p.Arg77His	2/19	-	0.0004	rs570176656,COSM294752,COSM538683,COSM1648717
42	SNV	chr10	43620336	G	A	194	108	86	0.443	RET	ENSG00000165731	NON_SYNONYMOUS_CODING	ENST00000340058.5:c.2945G>A	ENSP00000344798.4:p.Arg982His	18/19	-	-	rs368550200,COSM1264016
42	SNV	chr10	43620336	G	A	194	108	86	0.443	RET	ENSG00000165731	NON_SYNONYMOUS_CODING	ENST00000355710.3:c.2945G>A	ENSP00000347942.3:p.Arg982His	18/20	YES	-	rs368550200,COSM1264016
10	SNV	chr17	78345721	A	T	402	227	175	0.434	RNF213	ENSG00000173821	NON_SYNONYMOUS_CODING	ENST00000336301.6:c.6692A>T	ENSP00000338218.6:p.Lys2231Ile	22/43	-	-	-
10	SNV	chr17	78345721	A	T	402	227	175	0.434	RNF213	ENSG00000173821	NON_SYNONYMOUS_CODING	ENST00000508628.2:c.12620A>T	ENSP00000425956.2:p.Lys4207Ile	48/69	-	-	-
S32	SNV	chr17	78252690	C	T	612	323	289	0.472	RNF213	ENSG00000173821	NON_SYNONYMOUS_CODING	ENST00000508628.2:c.389C>T	ENSP00000425956.2:p.Pro130Leu	4/69	-	-	rs529986297
S52	SNV	chr17	78252690	C	T	698	361	337	0.483	RNF213	ENSG00000173821	NON_SYNONYMOUS_CODING	ENST00000508628.2:c.389C>T	ENSP00000425956.2:p.Pro130Leu	4/69	-	-	rs529986297
44	SNV	chr6	117609821	C	T	349	239	110	0.315	ROS1	ENSG00000047936	NON_SYNONYMOUS_CODING	ENST00000368508.3:c.6878G>A	ENSP00000357494.3:p.Gly2293Asp	43/43	YES	-	-
44	SNV	chr6	117609821	C	T	349	239	110	0.315	ROS1	ENSG00000047936	NON_SYNONYMOUS_CODING	ENST00000368507.3:c.6860G>A	ENSP00000357493.3:p.Gly2287Asp	44/44	-	-	-
S33	SNV	chr6	117622266	G	A	931	647	284	0.305	ROS1	ENSG00000047936	NON_SYNONYMOUS_CODING	ENST00000368508.3:c.6604C>T	ENSP00000357494.3:p.Pro2202Ser	42/43	YES	-	-
S33	SNV	chr6	117622266	G	A	931	647	284	0.305	ROS1	ENSG00000047936	NON_SYNONYMOUS_CODING	ENST00000368507.3:c.6586C>T	ENSP00000357493.3:p.Pro2196Ser	43/44	-	-	-
29	SNV	chr21	36171751	G	A	458	309	149	0.325	RUNX1	ENSG00000159216	STOP_GAINED	ENST00000344691.4:c.733C>T	ENSP00000340690.4:p.Gln245Ter	5/6	-	-	-
29	SNV	chr21	36171751	G	A	458	309	149	0.325	RUNX1	ENSG00000159216	STOP_GAINED	ENST00000300305.3:c.814C>T	ENSP00000300305.3:p.Gln272Ter	7/8	YES	-	-
29	SNV	chr21	36171751	G	A	458	309	149	0.325	RUNX1	ENSG00000159216	STOP_GAINED	ENST00000399240.1:c.541C>T	ENSP00000382184.1:p.Gln181Ter	4/5	-	-	-
29	SNV	chr21	36171751	G	A	458	309	149	0.325	RUNX1	ENSG00000159216	STOP_GAINED	ENST00000325074.5:c.778C>T	ENSP00000319459.5:p.Gln260Ter	6/7	-	-	-
29	SNV	chr21	36171751	G	A	458	309	149	0.325	RUNX1	ENSG00000159216	STOP_GAINED	ENST00000437180.1:c.814C>T	ENSP00000409227.1:p.Gln272Ter	8/9	-	-	-
35	SNV	chr21	36259178	G	A	165	71	94	0.57	RUNX1	ENSG00000159216	NON_SYNONYMOUS_CODING	ENST00000344691.4:c.232C>T	ENSP00000340690.4:p.His78Tyr	1/6	-	-	-
35	SNV	chr21	36259178	G	A	165	71	94	0.57	RUNX1	ENSG00000159216	NON_SYNONYMOUS_CODING	ENST00000437180.1:c.313C>T	ENSP00000409227.1:p.His105Tyr	4/9	-	-	-
35	SNV	chr21	36259178	G	A	165	71	94	0.57	RUNX1	ENSG00000159216	NON_SYNONYMOUS_CODING	ENST00000486278.2:c.241C>T	ENSP00000438019.1:p.His81Tyr	4/7	-	-	-
35	SNV	chr21	36259178	G	A	165	71	94	0.57	RUNX1	ENSG00000159216	NON_SYNONYMOUS_CODING	ENST00000325074.5:c.277C>T	ENSP00000319459.5:p.His93Tyr	2/7	-	-	-
35	SNV	chr21	36259178	G	A	165	71	94	0.57	RUNX1	ENSG00000159216	NON_SYNONYMOUS_CODING	ENST00000358356.5:c.232C>T	ENSP00000351123.5:p.His78Tyr	1/5	-	-	-
35	SNV	chr21	36259178	G	A	165	71	94	0.57	RUNX1	ENSG00000159216	NON_SYNONYMOUS_CODING	ENST00000399240.1:c.232C>T	ENSP00000382184.1:p.His78Tyr	1/5	-	-	-
35	SNV	chr21	36259178	G	A	165	71	94	0.57	RUNX1	ENSG00000159216	NON_SYNONYMOUS_CODING	ENST00000300305.3:c.313C>T	ENSP00000300305.3:p.His105Tyr	3/8	YES	-	-
35	SNV	chr21	36259178	G	A	165	71	94	0.57	RUNX1	ENSG00000159216	NON_SYNONYMOUS_CODING	ENST00000399237.2:c.277C>T	ENSP00000382182.2:p.His93Tyr	2/5	-	-	-
35	SNV	chr21	36259178	G	A	165	71	94	0.57	RUNX1	ENSG00000159216	NON_SYNONYMOUS_CODING	ENST00000455571.1:c.274C>T	ENSP00000388189.1:p.His92Tyr	3/4	-	-	-
43	SNV	chr21	36206822	C	A	127	67	60	0.472	RUNX1	ENSG00000159216	NON_SYNONYMOUS_CODING	ENST00000300305.3:c.690G>T	ENSP00000300305.3:p.Gln230His	6/8	YES	-	-
43	SNV	chr21	36206817	C	A	126	84	42	0.333	RUNX1	ENSG00000159216	NON_SYNONYMOUS_CODING	ENST00000300305.3:c.695G>T	ENSP00000300305.3:p.Arg232Leu	6/8	YES	-	rs368711448,COSM1030460
43	SNV	chr21	36206822	C	A	127	67	60	0.472	RUNX1	ENSG00000159216	NON_SYNONYMOUS_CODING	ENST00000325074.5:c.654G>T	ENSP00000319459.5:p.Gln218His	5/7	-	-	-
43	SNV	chr21	36206817	C	A	126	84	42	0.333	RUNX1	ENSG00000159216	NON_SYNONYMOUS_CODING	ENST00000358356.5:c.614G>T	ENSP00000351123.5:p.Arg205Leu	4/5	-	-	rs368711448,COSM1030460
43	INDEL	chr21	36206820	A	-G	126	67	59	0.468	RUNX1	ENSG00000159216	FRAME_SHIFT	ENST00000325074.5:c.656delT	ENSP00000319459.5:p.Leu219ArgfsTer6	5/7	-	-	-
43	SNV	chr21	36206817	C	A	126	84	42	0.333	RUNX1	ENSG00000159216	NON_SYNONYMOUS_CODING	ENST00000325074.5:c.659G>T	ENSP00000319459.5:p.Arg220Leu	5/7	-	-	rs368711448,COSM1030460
43	SNV	chr21	36206822	C	A	127	67	60	0.472	RUNX1	ENSG00000159216	NON_SYNONYMOUS_CODING	ENST00000358356.5:c.659G>T	ENSP00000351123.5:p.Arg220Leu	4/5	-	-	rs368711448,COSM1030460
43	SNV	chr21	36206822	C	A	127	67	60	0.472	RUNX1	ENSG00000159216	NON_SYNONYMOUS_CODING	ENST00000399237.2:c.654G>T	ENSP00000382182.2:p.Gln218His	5/5	-	-	-
43	SNV	chr21	36206822	C	A	127	67	60	0.472	RUNX1	ENSG00000159216	NON_SYNONYMOUS_CODING	ENST00000437180.1:c.690G>T	ENSP00000409227.1:p.Gln230His	7/9	-	-	-
43	SNV	chr21	36206817	C	A	126	84	42	0.333	RUNX1	ENSG00000159216	NON_SYNONYMOUS_CODING	ENST00000437180.1:c.695G>T	ENSP00000409227.1:p.Arg232Leu	7/9	-	-	rs368711448,COSM1030460
43	INDEL	chr21	36206820	A	-G	126	67	59	0.468	RUNX1	ENSG00000159216	FRAME_SHIFT	ENST00000344691.4:c.611delT	ENSP00000340690.4:p.Leu204ArgfsTer6	4/6	-	-	-
43	INDEL	chr21	36206820	A	-G	126	67	59	0.468	RUNX1	ENSG00000159216	FRAME_SHIFT	ENST00000358356.5:c.611delT	ENSP00000351123.5:p.Leu204ArgfsTer6	4/5	-	-	-
43	INDEL	chr21	36206820	A	-G	126	67	59	0.468	RUNX1	ENSG00000159216	FRAME_SHIFT	ENST00000399237.2:c.656delT	ENSP00000382182.2:p.Leu219ArgfsTer6	5/5	-	-	-
44	SNV	chr21	36259156	A	G	175	126	49	0.28	RUNX1	ENSG00000159216	NON_SYNONYMOUS_CODING	ENST00000399237.2:c.299T>C	ENSP00000382182.2:p.Leu100Pro	2/5	-	-	COSM3737026
44	SNV	chr21	36259156	A	G	175	126	49	0.28	RUNX1	ENSG00000159216	NON_SYNONYMOUS_CODING	ENST00000358356.5:c.254T>C	ENSP00000351123.5:p.Leu85Pro	1/5	-	-	COSM3737026
44	SNV	chr21	36259156	A	G	175	126	49	0.28	RUNX1	ENSG00000159216	NON_SYNONYMOUS_CODING	ENST00000437180.1:c.335T>C	ENSP00000409227.1:p.Leu112Pro	4/9	-	-	COSM3737026
44	SNV	chr21	36259156	A	G	175	126	49	0.28	RUNX1	ENSG00000159216	NON_SYNONYMOUS_CODING	ENST00000325074.5:c.299T>C	ENSP00000319459.5:p.Leu100Pro	2/7	-	-	COSM3737026
44	SNV	chr21	36259156	A	G	175	126	49	0.28	RUNX1	ENSG00000159216	NON_SYNONYMOUS_CODING	ENST00000300305.3:c.335T>C	ENSP00000300305.3:p.Leu112Pro	3/8	YES	-	COSM3737026
44	SNV	chr21	36259156	A	G	175	126	49	0.28	RUNX1	ENSG00000159216	NON_SYNONYMOUS_CODING	ENST00000399240.1:c.254T>C	ENSP00000382184.1:p.Leu85Pro	1/5	-	-	COSM3737026
44	SNV	chr21	36259156	A	G	175	126	49	0.28	RUNX1	ENSG00000159216	NON_SYNONYMOUS_CODING	ENST00000344691.4:c.254T>C	ENSP00000340690.4:p.Leu85Pro	1/6	-	-	COSM3737026
44	SNV	chr21	36259156	A	G	175	126	49	0.28	RUNX1	ENSG00000159216	NON_SYNONYMOUS_CODING	ENST00000486278.2:c.263T>C	ENSP00000438019.1:p.Leu88Pro	4/7	-	-	COSM3737026
44	SNV	chr21	36259156	A	G	175	126	49	0.28	RUNX1	ENSG00000159216	NON_SYNONYMOUS_CODING	ENST00000455571.1:c.296T>C	ENSP00000388189.1:p.Leu99Pro	3/4	-	-	COSM3737026
6	SNV	chr21	36231773	C	T	664	65	599	0.901	RUNX1	ENSG00000159216	NON_SYNONYMOUS_CODING	ENST00000399240.1:c.530G>A	ENSP00000382184.1:p.Arg177Gln	3/5	-	-	COSM24731,COSM1413953,COSM3423938,CM093278
6	SNV	chr21	36231773	C	T	664	65	599	0.901	RUNX1	ENSG00000159216	NON_SYNONYMOUS_CODING	ENST00000399237.2:c.575G>A	ENSP00000382182.2:p.Arg192Gln	4/5	-	-	COSM24731,COSM1413953,COSM3423938,CM093278

6	SNV	chr21	36231773	C	T	664	65	599	0.901	RUNX1	ENSG00000159216	NON_SYNONYMOUS_CODING	ENST00000486278.2:c.539G>A	ENSP00000438019.1:p.Arg180Gln	6/7	-	-	COSM24731,COSM1413953,COSM3423938,CM093278
6	SNV	chr21	36231773	C	T	664	65	599	0.901	RUNX1	ENSG00000159216	NON_SYNONYMOUS_CODING	ENST00000358356.5:c.530G>A	ENSP00000351123.5:p.Arg177Gln	3/5	-	-	COSM24731,COSM1413953,COSM3423938,CM093278
6	SNV	chr21	36231773	C	T	664	65	599	0.901	RUNX1	ENSG00000159216	NON_SYNONYMOUS_CODING	ENST00000344691.4:c.530G>A	ENSP00000340690.4:p.Arg177Gln	3/6	-	-	COSM24731,COSM1413953,COSM3423938,CM093278
6	SNV	chr21	36231773	C	T	664	65	599	0.901	RUNX1	ENSG00000159216	NON_SYNONYMOUS_CODING	ENST00000300305.3:c.611G>A	ENSP00000300305.3:p.Arg204Gln	5/8	YES	-	COSM24731,COSM1413953,COSM3423938,CM093278
6	SNV	chr21	36231773	C	T	664	65	599	0.901	RUNX1	ENSG00000159216	NON_SYNONYMOUS_CODING	ENST00000325074.5:c.575G>A	ENSP00000319459.5:p.Arg192Gln	4/7	-	-	COSM24731,COSM1413953,COSM3423938,CM093278
6	SNV	chr21	36231773	C	T	664	65	599	0.901	RUNX1	ENSG00000159216	NON_SYNONYMOUS_CODING	ENST00000437180.1:c.611G>A	ENSP00000409227.1:p.Arg204Gln	6/9	-	-	COSM24731,COSM1413953,COSM3423938,CM093278
62	SNV	chr21	36252880	A	C	181	129	52	0.286	RUNX1	ENSG00000159216	NON_SYNONYMOUS_CODING	ENST00000399237.2:c.446T>G	ENSP00000382182.2:p.Leu149Arg	3/5	-	-	COSM44417
62	SNV	chr21	36252880	A	C	181	129	52	0.286	RUNX1	ENSG00000159216	NON_SYNONYMOUS_CODING	ENST00000486278.2:c.410T>G	ENSP00000438019.1:p.Leu137Arg	5/7	-	-	COSM44417
62	SNV	chr21	36252880	A	C	181	129	52	0.286	RUNX1	ENSG00000159216	NON_SYNONYMOUS_CODING	ENST00000300305.3:c.482T>G	ENSP00000300305.3:p.Leu161Arg	4/8	YES	-	COSM44417
62	SNV	chr21	36252880	A	C	181	129	52	0.286	RUNX1	ENSG00000159216	NON_SYNONYMOUS_CODING	ENST00000437180.1:c.482T>G	ENSP00000409227.1:p.Leu161Arg	5/9	-	-	COSM44417
62	SNV	chr21	36252880	A	C	181	129	52	0.286	RUNX1	ENSG00000159216	NON_SYNONYMOUS_CODING	ENST00000344691.4:c.401T>G	ENSP00000340690.4:p.Leu134Arg	2/6	-	-	COSM44417
62	SNV	chr21	36252880	A	C	181	129	52	0.286	RUNX1	ENSG00000159216	NON_SYNONYMOUS_CODING	ENST00000325074.5:c.446T>G	ENSP00000319459.5:p.Leu149Arg	3/7	-	-	COSM44417
62	SNV	chr21	36252880	A	C	181	129	52	0.286	RUNX1	ENSG00000159216	NON_SYNONYMOUS_CODING	ENST00000399240.1:c.401T>G	ENSP00000382184.1:p.Leu134Arg	2/5	-	-	COSM44417
62	SNV	chr21	36252880	A	C	181	129	52	0.286	RUNX1	ENSG00000159216	NON_SYNONYMOUS_CODING	ENST00000358356.5:c.401T>G	ENSP00000351123.5:p.Leu134Arg	2/5	-	-	COSM44417
63	INDEL	chr21	36164652	C	+CGTA	118	63	55	0.458	RUNX1	ENSG00000159216	FRAME_SHIFT	-	-	9/9	-	-	-
63	INDEL	chr21	36164652	C	+CGTA	118	63	55	0.458	RUNX1	ENSG00000159216	FRAME_SHIFT	-	-	6/6	-	-	-
63	INDEL	chr21	36164652	C	+CGTA	118	63	55	0.458	RUNX1	ENSG00000159216	FRAME_SHIFT	-	-	7/7	-	-	-
63	INDEL	chr21	36164652	C	+CGTA	118	63	55	0.458	RUNX1	ENSG00000159216	FRAME_SHIFT	-	-	5/5	-	-	-
63	INDEL	chr21	36164652	C	+CGTA	118	63	55	0.458	RUNX1	ENSG00000159216	FRAME_SHIFT	-	-	8/8	YES	-	-
S03	SNV	chr21	36231782	C	T	437	205	232	0.531	RUNX1	ENSG00000159216	NON_SYNONYMOUS_CODING	ENST00000399237.2:c.566G>A	ENSP00000382182.2:p.Arg189Gln	4/5	-	-	rs74315450,COSM24805,CM992140
S03	SNV	chr21	36231782	C	T	437	205	232	0.531	RUNX1	ENSG00000159216	NON_SYNONYMOUS_CODING	ENST00000399240.1:c.521G>A	ENSP00000382184.1:p.Arg174Gln	3/5	-	-	rs74315450,COSM24805,CM992140
S03	SNV	chr21	36259163	T	G	276	138	138	0.498	RUNX1	ENSG00000159216	NON_SYNONYMOUS_CODING	ENST00000437180.1:c.328A>C	ENSP00000409227.1:p.Lys110Gln	4/9	-	-	rs121912498,COSM4170263,CM025982
S03	SNV	chr21	36231782	C	T	437	205	232	0.531	RUNX1	ENSG00000159216	NON_SYNONYMOUS_CODING	ENST00000437180.1:c.602G>A	ENSP00000409227.1:p.Arg201Gln	6/9	-	-	rs74315450,COSM24805,CM992140
S03	SNV	chr21	36259163	T	G	276	138	138	0.498	RUNX1	ENSG00000159216	NON_SYNONYMOUS_CODING	ENST00000455571.1:c.289A>C	ENSP00000388189.1:p.Lys97Gln	3/4	-	-	rs121912498,COSM4170263,CM025982
S03	SNV	chr21	36259163	T	G	276	138	138	0.498	RUNX1	ENSG00000159216	NON_SYNONYMOUS_CODING	ENST00000344691.4:c.247A>C	ENSP00000340690.4:p.Lys83Gln	1/6	-	-	rs121912498,COSM4170263,CM025982
S03	SNV	chr21	36259163	T	G	276	138	138	0.498	RUNX1	ENSG00000159216	NON_SYNONYMOUS_CODING	ENST00000358356.5:c.247A>C	ENSP00000351123.5:p.Lys83Gln	1/5	-	-	rs121912498,COSM4170263,CM025982
S03	SNV	chr21	36259163	T	G	276	138	138	0.498	RUNX1	ENSG00000159216	NON_SYNONYMOUS_CODING	ENST00000399240.1:c.247A>C	ENSP00000382184.1:p.Lys83Gln	1/5	-	-	rs121912498,COSM4170263,CM025982
S03	SNV	chr21	36231782	C	T	437	205	232	0.531	RUNX1	ENSG00000159216	NON_SYNONYMOUS_CODING	ENST00000486278.2:c.530G>A	ENSP00000438019.1:p.Arg177Gln	6/7	-	-	rs74315450,COSM24805,CM992140
S03	SNV	chr21	36259163	T	G	276	138	138	0.498	RUNX1	ENSG00000159216	NON_SYNONYMOUS_CODING	ENST00000399237.2:c.292A>C	ENSP00000382182.2:p.Lys98Gln	2/5	-	-	rs121912498,COSM4170263,CM025982
S03	SNV	chr21	36259163	T	G	276	138	138	0.498	RUNX1	ENSG00000159216	NON_SYNONYMOUS_CODING	ENST00000300305.3:c.328A>C	ENSP00000300305.3:p.Lys110Gln	3/8	YES	-	rs121912498,COSM4170263,CM025982
S03	SNV	chr21	36231782	C	T	437	205	232	0.531	RUNX1	ENSG00000159216	NON_SYNONYMOUS_CODING	ENST00000300305.3:c.602G>A	ENSP00000300305.3:p.Arg201Gln	5/8	YES	-	rs74315450,COSM24805,CM992140
S03	SNV	chr21	36231782	C	T	437	205	232	0.531	RUNX1	ENSG00000159216	NON_SYNONYMOUS_CODING	ENST00000325074.5:c.566G>A	ENSP00000319459.5:p.Arg189Gln	4/7	-	-	rs74315450,COSM24805,CM992140
S03	SNV	chr21	36259163	T	G	276	138	138	0.498	RUNX1	ENSG00000159216	NON_SYNONYMOUS_CODING	ENST00000325074.5:c.292A>C	ENSP00000319459.5:p.Lys98Gln	2/7	-	-	rs121912498,COSM4170263,CM025982
S03	SNV	chr21	36259163	T	G	276	138	138	0.498	RUNX1	ENSG00000159216	NON_SYNONYMOUS_CODING	ENST00000486278.2:c.256A>C	ENSP00000438019.1:p.Lys86Gln	4/7	-	-	rs121912498,COSM4170263,CM025982
S03	SNV	chr21	36231782	C	T	437	205	232	0.531	RUNX1	ENSG00000159216	NON_SYNONYMOUS_CODING	ENST00000344691.4:c.521G>A	ENSP00000340690.4:p.Arg174Gln	3/6	-	-	rs74315450,COSM24805,CM992140
S03	SNV	chr21	36231782	C	T	437	205	232	0.531	RUNX1	ENSG00000159216	NON_SYNONYMOUS_CODING	ENST00000358356.5:c.521G>A	ENSP00000351123.5:p.Arg174Gln	3/5	-	-	rs74315450,COSM24805,CM992140
S17	SNV	chr21	36259172	G	T	324	196	128	0.395	RUNX1	ENSG00000159216	NON_SYNONYMOUS_CODING	ENST00000300305.3:c.319C>A	ENSP00000300305.3:p.Arg107Ser	3/8	YES	-	COSM24736
S17	SNV	chr21	36259172	G	T	324	196	128	0.395	RUNX1	ENSG00000159216	NON_SYNONYMOUS_CODING	ENST00000358356.5:c.238C>A	ENSP00000351123.5:p.Arg80Ser	1/5	-	-	COSM24736
S17	SNV	chr21	36259172	G	T	324	196	128	0.395	RUNX1	ENSG00000159216	NON_SYNONYMOUS_CODING	ENST00000344691.4:c.238C>A	ENSP00000340690.4:p.Arg80Ser	1/6	-	-	COSM24736
S17	SNV	chr21	36259172	G	T	324	196	128	0.395	RUNX1	ENSG00000159216	NON_SYNONYMOUS_CODING	ENST00000455571.1:c.280C>A	ENSP00000388189.1:p.Arg94Ser	3/4	-	-	COSM24736
S17	SNV	chr21	36259172	G	T	324	196	128	0.395	RUNX1	ENSG00000159216	NON_SYNONYMOUS_CODING	ENST00000325074.5:c.283C>A	ENSP00000319459.5:p.Arg95Ser	2/7	-	-	COSM24736
S17	SNV	chr21	36259172	G	T	324	196	128	0.395	RUNX1	ENSG00000159216	NON_SYNONYMOUS_CODING	ENST00000399240.1:c.238C>A	ENSP00000382184.1:p.Arg80Ser	1/5	-	-	COSM24736
S17	SNV	chr21	36259172	G	T	324	196	128	0.395	RUNX1	ENSG00000159216	NON_SYNONYMOUS_CODING	ENST00000486278.2:c.247C>A	ENSP00000438019.1:p.Arg83Ser	4/7	-	-	COSM24736
S17	SNV	chr21	36259172	G	T	324	196	128	0.395	RUNX1	ENSG00000159216	NON_SYNONYMOUS_CODING	ENST00000437180.1:c.319C>A	ENSP00000409227.1:p.Arg107Ser	4/9	-	-	COSM24736
S17	SNV	chr21	36259172	G	T	324	196	128	0.395	RUNX1	ENSG00000159216	NON_SYNONYMOUS_CODING	ENST00000399237.2:c.283C>A	ENSP00000382182.2:p.Arg95Ser	2/5	-	-	COSM24736
S21	INDEL	chr21	36253000	A	+G	468	267	201	0.43	RUNX1	ENSG00000159216	FRAME_SHIFT	-	-	5/7	-	-	-
S21	INDEL	chr21	36253000	A	+G	468	267	201	0.43	RUNX1	ENSG00000159216	FRAME_SHIFT	-	-	2/5	-	-	-
S21	INDEL	chr21	36253000	A	+G	468	267	201	0.43	RUNX1	ENSG00000159216	FRAME_SHIFT	-	-	3/5	-	-	-
S21	INDEL	chr21	36253000	A	+G	468	267	201	0.43	RUNX1	ENSG00000159216	FRAME_SHIFT	-	-	5/9	-	-	-
S21	INDEL	chr21	36253000	A	+G	468	267	201	0.43	RUNX1	ENSG00000159216	FRAME_SHIFT	-	-	3/7	-	-	-
S21	INDEL	chr21	36253000	A	+G	468	267	201	0.43	RUNX1	ENSG00000159216	FRAME_SHIFT	-	-	4/8	YES	-	-
S21	INDEL	chr21	36253000	A	+G	468	267	201	0.43	RUNX1	ENSG00000159216	FRAME_SHIFT	-	-	2/6	-	-	-
S21	INDEL	chr21	36253000	A	+G	468	267	201	0.43	RUNX1	ENSG00000159216	FRAME_SHIFT	-	-	2/5	-	-	-
S21	INDEL	chr21	36253000	A	+G	468	267	201	0.43	RUNX1	ENSG00000159216	FRAME_SHIFT	-	-	4/4	-	-	-
S34	SNV	chr21	36259172	G	A	366	113	253	0.691	RUNX1	ENSG00000159216	NON_SYNONYMOUS_CODING	ENST00000399237.2:c.283C>T	ENSP00000382182.2:p.Arg95Cys	2/5	-	-	COSM24736
S34	SNV	chr21	36259172	G	A	366	113	253	0.691	RUNX1	ENSG00000159216	NON_SYNONYMOUS_CODING	ENST00000300305.3:c.319C>T	ENSP00000300305.3:p.Arg107Cys	3/8	YES	-	COSM24736
S34	SNV	chr21	36259172	G	A	366	113	253	0.691	RUNX1	ENSG00000159216	NON_SYNONYMOUS_CODING	ENST00000344691.4:c.238C>T	ENSP00000340690.4:p.Arg80Cys	1/6	-	-	COSM24736
S34	SNV	chr21	36259172	G	A	366	113	253	0.691	RUNX1	ENSG00000159216	NON_SYNONYMOUS_CODING	ENST00000437180.1:c.319C>T	ENSP00000409227.1:p.Arg107Cys	4/9	-	-	COSM24736
S34	SNV	chr21	36259172	G	A	366	113	253	0.691	RUNX1	ENSG00000159216	NON_SYNONYMOUS_CODING	ENST00000455571.1:c.280C>T	ENSP00000388189.1:p.Arg94Cys	3/4	-	-	COSM24736
S34	SNV	chr21	36259172	G	A	366	113	253	0.691	RUNX1	ENSG00000159216	NON_SYNONYMOUS_CODING	ENST00000486278.2:c.247C>T	ENSP00000438019.1:p.Arg83Cys	4/7	-	-	COSM24736
S34	SNV	chr21	36259172	G	A	366	113	253	0.691	RUNX1	ENSG00000159216	NON_SYNONYMOUS_CODING	ENST00000325074.5:c.283C>T	ENSP00000319459.5:p.Arg95Cys	2/7	-	-	COSM24736
S34	SNV	chr21	36259172	G	A	366	113	253	0.691	RUNX1	ENSG00000159216	NON_SYNONYMOUS_CODING	ENST00000358356.5:c.238C>T	ENSP00000351123.5:p.Arg80Cys	1/5	-	-	COSM24736
S34	SNV	chr21	36259172	G	A	366	113	253	0.691	RUNX1	ENSG00000159216	NON_SYNONYMOUS_CODING	ENST00000399240.1:c.238C>T	ENSP00000382184.1:p.Arg80Cys	1/5	-	-	COSM24736
S35	INDEL	chr21	36164771	C	-ATGCCGATGCCGAT	148	95	53	0.358	RUNX1	ENSG00000159216	FRAME_SHIFT	ENST00000437180.1:c.1104delG	ENSP00000409227.1:p.Met368IlefsTer226	9/9	-	-	-
S35	INDEL	chr21	36164771	C	-ATGCCGATGCCGAT	148	95	53	0.358	RUNX1	ENSG00000159216	FRAME_SHIFT	ENST00000300305.3:c.1104delG	ENSP00000300305.3:p.Met368IlefsTer226	8/8	YES	-	-
S35	INDEL	chr21	36164771	C	-ATGCCGATGCCGAT	148	95	53	0.358	RUNX1	ENSG00000159216	FRAME_SHIFT	ENST00000399240.1:c.831delG	ENSP00000382184.1:p.Met277IlefsTer226	5/5	-	-	-
S35	INDEL	chr21	36164771	C	-ATGCCGATGCCGAT	148	95	53	0.358	RUNX1	ENSG00000159216	FRAME_SHIFT	ENST00000325074.5:c.1068delG	ENSP00000319459.5:p.Met356IlefsTer226	7/7	-	-	-
S35	INDEL	chr21	36164771	C	-ATGCCGATGCCGAT	148	95	53	0.358	RUNX1	ENSG00000159216	FRAME_SHIFT	ENST00000344691.4:c.1023delG	ENSP00000340690.4:p.Met341IlefsTer226	6/6	-	-	-
S36	SNV	chr21	36259156	A	G	361	214	147	0.407	RUNX1	ENSG00000159216	NON_SYNONYMOUS_CODING	ENST00000300305.3:c.335T>C	ENSP00000300305.3:p.Leu112Pro	3/8	YES	-	COSM3737026
S36	SNV	chr21	36259156	A	G	361	214	147	0.407	RUNX1	ENSG00000159216	NON_SYNONYMOUS_CODING	ENST00000486278.2:c.263T>C	ENSP00000438019.1:p.Leu88Pro	4/7	-	-	COSM3737026
S36	SNV	chr21	36259156	A	G	361	214	147	0.407	RUNX1	ENSG00000159216	NON_SYNONYMOUS_CODING	ENST00000325074.5:c.299T>C	ENSP00000319459.5:p.Leu100Pro	2/7	-	-	COSM3737026
S36	SNV	chr21	36259156	A	G	361	214	147	0.407	RUNX1	ENSG00000159216	NON_SYNONYMOUS_CODING	ENST00000437180.1:c.335T>C	ENSP00000409227.1:p.Leu112Pro	4/9	-	-	COSM3737026
S36	SNV	chr21	36259156	A	G	361	214	147	0.407	RUNX1	ENSG00000159216	NON_SYNONYMOUS_CODING	ENST00000344691.4:c.254T>C	ENSP00000340690.4:p.Leu85Pro	1/6	-	-	COSM3737026
S36	SNV	chr21	36259156	A	G	361	214	147	0.407	RUNX1	ENSG00000159216	NON_SYNONYMOUS_CODING	ENST00000455571.1:c.296T>C	ENSP00000388189.1:p.Leu99Pro	3/4	-	-	COSM3737026
S36	SNV	chr21	36259156	A	G	361	214	147	0.407	RUNX1	ENSG00000159216	NON_SYNONYMOUS_CODING	ENST00000399240.1:c.254T>C	ENSP00000382184.1:p.Leu85Pro	1/5	-	-	COSM3737026
S36	SNV	chr21	36259156	A	G	361	214	147	0.407	RUNX1	ENSG00000159216	NON_SYNONYMOUS_CODING	ENST00000358356.5:c.254T>C	ENSP00000351123.5:p.Leu85Pro	1/5	-	-	COSM3737026
S36	SNV	chr21	36259156	A	G	361	214	147	0.407	RUNX1	ENSG00000159216	NON_SYNONYMOUS_CODING	ENST00000399237.2:c.299T>C	ENSP00000382182.2:p.Leu100Pro	2/5	-	-	COSM3737026
S43	SNV	chr21	36231783	G	A	500	275	225	0.45	RUNX1	ENSG00000159216	STOP_GAINED	ENST00000344691.4:c.520C>T	ENSP00000340690.4:p.Arg174Ter	3/6	-	-	COSM186279,COSM24771,COSM3356082,CM992141
S43	SNV	chr21	36231783	G	A	500	275	225	0.45	RUNX1	ENSG00000159216	STOP_GAINED	ENST00000325074.5:c.565C>T	ENSP00000319459.5:p.Arg189Ter	4/7	-	-	COSM186279,COSM24771,COSM3356082,CM992141
S43	SNV	chr21	36231783	G	A	500	275	225	0.45	RUNX1	ENSG00000159216	STOP_GAINED	ENST00000300305.3:c.601C>T	ENSP00000300305.3:p.Arg201Ter	5/8	YES	-	COSM186279,COSM24771,COSM3356082,CM992141
S43	SNV	chr21	36231783	G	A	500	275	225	0.45	RUNX1	ENSG00000159216	STOP_GAINED	ENST00000437180.1:c.601C>T	ENSP00000409227.1:p.Arg201Ter	6/9	-	-	COSM186279,COSM24771,COSM3356082,CM992141
S43	SNV	chr21	36231783	G	A	500	275	225	0.45	RUNX1	ENSG00000159216	STOP_GAINED	ENST00000399240.1:c.520C>T	ENSP00000382184.1:p.Arg174Ter	3/5	-	-	COSM186279,COSM24771,COSM3356082,CM992141
S43	SNV	chr21	36231783	G	A	500	275	225	0.45	RUNX1	ENSG00000159216	STOP_GAINED	ENST00000486278.2:c.529C>T	ENSP00000438019.1:p.Arg177Ter	6/7	-	-	COSM186279,COSM24771,COSM3356082,CM992141
S43	SNV	chr21	36231783	G	A	500	275	225	0.45	RUNX1	ENSG00000159216	STOP_GAINED	ENST00000399237.2:c.565C>T	ENSP00000382182.2:p.Arg189Ter	4/5	-	-	COSM186279,COSM24771,COSM3356082,CM992141
S43	SNV	chr21	36231783	G	A	500	275	225	0.45	RUNX1	ENSG00000159216	STOP_GAINED	ENST00000358356.5:c.520C>T	ENSP00000351123.5:p.Arg174Ter	3/5	-	-	COSM186279,COSM24771,COSM3356082,CM992141
S53	SNV	chr21	36252877	C	T	575	336	239	0.415	RUNX1	ENSG00000159216	NON_SYNONYMOUS_CODING	ENST00000399237.2:c.449G>A	ENSP00000382182.2:p.Arg150Lys	3/5	-	-	COSM96546
S53	SNV	chr21	36252877	C	T	575	336	239	0.415	RUNX1	ENSG00000159216	NON_SYNONYMOUS_CODING	ENST00000325074.5:c.449G>A	ENSP00000319459.5:p.Arg150Lys	3/7	-	-	COSM96546
S53	SNV	chr21	36252877	C	T	575	336	239	0.415	RUNX1	ENSG00000159216	NON_SYNONYMOUS_CODING	ENST00000399240.1:c.404G>A	ENSP00000382184.1:p.Arg135Lys	2/5	-	-	COSM96546
S53	SNV	chr21	36252877	C	T	575	336	239	0.415	RUNX1	ENSG00000159216	NON_SYNONYMOUS_CODING	ENST00000344691.4:c.404G>A	ENSP00000340690.4:p.Arg135Lys	2/6	-	-	COSM96546
S53	SNV	chr21	36252877	C	T	575	336	239	0.415	RUNX1	ENSG00000159216	NON_SYNONYMOUS_CODING	ENST00000486278.2:c.413G>A	ENSP00000438019.1:p.Arg138Lys	5/7	-	-	COSM96546
S53	SNV	chr21	36252877	C	T	575	336	239	0.415	RUNX1	ENSG00000159216	NON_SYNONYMOUS_CODING	ENST00000358356.5:c.404G>A	ENSP00000351123.5:p.Arg135Lys	2/5	-	-	COSM96546
S53	SNV	chr21	36252877	C	T	575	336	239	0.415	RUNX1	ENSG00000159216	NON_SYNONYMOUS_CODING	ENST00000300305.3:c.485G>A	ENSP00000300305.3:p.Arg162Lys	4/8	YES	-	COSM96546
S53	SNV	chr21	36252877	C	T	575	336	239	0.415	RUNX1	ENSG00000159216	NON_SYNONYMOUS_CODING	ENST00000437180.1:c.485G>A	ENSP00000409227.1:p.Arg162Lys	5/9	-	-	COSM96546
S56	SNV	chr21	36171607	G	A	1080	638	442	0.409	RUNX1	ENSG00000159216	STOP_GAINED	ENST00000344691.4:c.877C>T	ENSP00000340690.4:p.Arg293Ter	5/6	-	-	COSM41699,COSM1413949,CM086912
S56	SNV	chr21	36171607	G	A	1080	638	442	0.409	RUNX1	ENSG00000159216	STOP_GAINED	ENST00000325074.5:c.922C>T	ENSP00000319459.5:p.Arg308Ter	6/7	-	-	COSM41699,COSM1413949,CM086912
S56	SNV	chr21	36171607	G	A	1080	638	442	0.409	RUNX1	ENSG00000159216	STOP_GAINED	ENST00000399240.1:c.685C>T	ENSP00000382184.1:p.Arg229Ter	4/5	-	-	COSM41699,COSM1413949,CM086912
S56	SNV	chr21	36171607	G	A	1080	638	442	0.409	RUNX1	ENSG00000159216	STOP_GAINED	ENST00000300305.3:c.958C>T	ENSP00000300305.3:p.Arg320Ter	7/8	YES	-	COSM41699,COSM1413949,CM086912
S56	SNV	chr21	36171607	G	A	1080	638	442	0.409	RUNX1	ENSG00000159216	STOP_GAINED	ENST00000437180.1:c.958C>T	ENSP00000409227.1:p.Arg320Ter	8/9	-	-	COSM41699,COSM1413949,CM086912
1	INDEL	chr10	102107196	G	-CGACCC	61	40	21	0.344	SCD	ENSG00000099194	UTR_5_PRIME	ENST00000370355.2:c.-64delG	-	1/6	YES	-	-

33	SNV	chr1	17380536	C	G	116	66	50	0.431	SDHB	ENSG00000117118	UTR_5_PRIME	ENST00000375499.3:c.-22G>C	-	1/8	YES	-	-	
37	SNV	chr11	111957665	G	A	44	20	24	0.546	SDHD	ENSG00000204370	NON_SYNONYMOUS_CODING	ENST00000525291.1:c.34G>A	ENSP00000436669.1:p.Gly12Ser	1/3	-	0.0050	rs34677591,CM003169	
37	SNV	chr11	111957665	G	A	44	20	24	0.546	SDHD	ENSG00000204370	NON_SYNONYMOUS_CODING	ENST00000375549.3:c.34G>A	ENSP00000364699.3:p.Gly12Ser	1/4	YES	0.0050	rs34677591,CM003169	
37	SNV	chr11	111957665	G	A	44	20	24	0.546	SDHD	ENSG00000204370	NON_SYNONYMOUS_CODING	ENST00000528182.1:c.34G>A	ENSP00000435475.1:p.Gly12Ser	1/4	-	0.0050	rs34677591,CM003169	
37	SNV	chr11	111957665	G	A	44	20	24	0.546	SDHD	ENSG00000204370	NON_SYNONYMOUS_CODING	ENST00000528021.1:c.34G>A	ENSP00000432465.1:p.Gly12Ser	1/4	-	0.0050	rs34677591,CM003169	
37	SNV	chr11	111957665	G	A	44	20	24	0.546	SDHD	ENSG00000204370	NON_SYNONYMOUS_CODING	ENST00000526592.1:c.34G>A	ENSP00000432005.1:p.Gly12Ser	1/5	-	0.0050	rs34677591,CM003169	
37	SNV	chr11	111957665	G	A	44	20	24	0.546	SDHD	ENSG00000204370	NON_SYNONYMOUS_CODING	ENST00000528048.1:c.34G>A	ENSP00000436217.1:p.Gly12Ser	1/3	-	0.0050	rs34677591,CM003169	
S25	SNV	chr11	111958677	A	G	1238	98	1140	0.921	SDHD	ENSG00000204370	NON_SYNONYMOUS_CODING	ENST00000375549.3:c.149A>G	ENSP00000364699.3:p.His50Arg	2/4	YES	0.0066	rs11214077,CM022244	
S25	SNV	chr11	111958677	A	G	1238	98	1140	0.921	SDHD	ENSG00000204370	NON_SYNONYMOUS_CODING	ENST00000528021.1:c.149A>G	ENSP00000432465.1:p.His50Arg	2/4	-	0.0066	rs11214077,CM022244	
S25	SNV	chr11	111958677	A	G	1238	98	1140	0.921	SDHD	ENSG00000204370	NON_SYNONYMOUS_CODING	ENST00000528048.1:c.149A>G	ENSP00000436217.1:p.His50Arg	2/3	-	0.0066	rs11214077,CM022244	
S25	SNV	chr11	111958677	A	G	1238	98	1140	0.921	SDHD	ENSG00000204370	NON_SYNONYMOUS_CODING	ENST00000526592.1:c.149A>G	ENSP00000432005.1:p.His50Arg	2/5	-	0.0066	rs11214077,CM022244	
S25	SNV	chr11	111958677	A	G	1238	98	1140	0.921	SDHD	ENSG00000204370	NON_SYNONYMOUS_CODING	ENST00000528182.1:c.149A>G	ENSP00000435475.1:p.His50Arg	2/4	-	0.0066	rs11214077,CM022244	
6	SNV	chr2	242276931	G	A	760	450	310	0.407	SEPT2	ENSG00000168385	NON_SYNONYMOUS_CODING	ENST00000360051.3:c.476G>A	ENSP00000353157.3:p.Gly159Glu	7/14	-	-	-	
6	SNV	chr2	242276931	G	A	760	450	310	0.407	SEPT2	ENSG00000168385	NON_SYNONYMOUS_CODING	ENST00000401990.1:c.506G>A	ENSP00000385109.1:p.Gly169Glu	10/17	-	-	-	
6	SNV	chr2	242276931	G	A	760	450	310	0.407	SEPT2	ENSG00000168385	NON_SYNONYMOUS_CODING	ENST00000428282.1:c.356G>A	ENSP00000397195.1:p.Gly119Glu	5/7	-	-	-	
6	SNV	chr2	242276931	G	A	760	450	310	0.407	SEPT2	ENSG00000168385	NON_SYNONYMOUS_CODING	ENST00000421717.1:c.41G>A	ENSP00000408296.1:p.Gly14Glu	1/7	-	-	-	
6	SNV	chr2	242276931	G	A	760	450	310	0.407	SEPT2	ENSG00000168385	NON_SYNONYMOUS_CODING	ENST00000391973.2:c.476G>A	ENSP00000375834.2:p.Gly159Glu	6/13	YES	-	-	
6	SNV	chr2	242276931	G	A	760	450	310	0.407	SEPT2	ENSG00000168385	NON_SYNONYMOUS_CODING	ENST00000402092.2:c.476G>A	ENSP00000385172.2:p.Gly159Glu	7/14	-	-	-	
6	SNV	chr2	242276931	G	A	760	450	310	0.407	SEPT2	ENSG00000168385	NON_SYNONYMOUS_CODING	ENST00000449239.1:c.476G>A	ENSP00000391717.1:p.Gly159Glu	5/6	-	-	-	
6	SNV	chr2	242276931	G	A	760	450	310	0.407	SEPT2	ENSG00000168385	NON_SYNONYMOUS_CODING	ENST00000443492.1:c.356G>A	ENSP00000399195.1:p.Gly119Glu	6/7	-	-	-	
6	SNV	chr2	242276931	G	A	760	450	310	0.407	SEPT2	ENSG00000168385	NON_SYNONYMOUS_CODING	ENST00000407971.1:c.356G>A	ENSP00000384525.1:p.Gly119Glu	5/12	-	-	-	
6	SNV	chr2	242276931	G	A	760	450	310	0.407	SEPT2	ENSG00000168385	NON_SYNONYMOUS_CODING	ENST00000391971.2:c.476G>A	ENSP00000375832.2:p.Gly159Glu	6/13	-	-	-	
7	INDEL	chrX	118827099	T	+GCCGCCGCAACGGGA	67	43	24	0.358	SEPT6	ENSG00000125354	UTR_5_PRIME	-	-	1/11	-	-	-	
7	INDEL	chrX	118827099	T	+GCCGCCGCAACGGGA	67	43	24	0.358	SEPT6	ENSG00000125354	UTR_5_PRIME	-	-	1/11	-	-	-	
7	INDEL	chrX	118827099	T	+GCCGCCGCAACGGGA	67	43	24	0.358	SEPT6	ENSG00000125354	UTR_5_PRIME	-	-	1/10	-	-	-	
7	INDEL	chrX	118827099	T	+GCCGCCGCAACGGGA	67	43	24	0.358	SEPT6	ENSG00000125354	UTR_5_PRIME	-	-	1/10	-	-	-	
7	INDEL	chrX	118827099	T	+GCCGCCGCAACGGGA	67	43	24	0.358	SEPT6	ENSG00000125354	UTR_5_PRIME	-	-	1/10	YES	-	-	
7	INDEL	chrX	118827099	T	+GCCGCCGCAACGGGA	67	43	24	0.358	SEPT6	ENSG00000125354	UTR_5_PRIME	-	-	1/11	-	-	-	
7	INDEL	chrX	118827099	T	+GCCGCCGCAACGGGA	67	43	24	0.358	SEPT6	ENSG00000125354	UTR_5_PRIME	-	-	1/10	-	-	-	
1	SNV	chr17	75494755	A	C	55	44	11	0.2	SEPT9	ENSG00000184640	UTR_3_PRIME	ENST00000449803.2:c.*15A>C	-	12/12	-	-	-	
1	SNV	chr17	75494755	A	C	55	44	11	0.2	SEPT9	ENSG00000184640	UTR_3_PRIME	ENST00000427674.2:c.*15A>C	-	11/11	-	-	-	
1	SNV	chr17	75494755	A	C	55	44	11	0.2	SEPT9	ENSG00000184640	UTR_3_PRIME	ENST00000427177.1:c.*15A>C	-	12/12	YES	-	-	
1	SNV	chr17	75494755	A	C	55	44	11	0.2	SEPT9	ENSG00000184640	UTR_3_PRIME	ENST00000541152.2:c.*15A>C	-	10/10	-	-	-	
1	SNV	chr17	75494755	A	C	55	44	11	0.2	SEPT9	ENSG00000184640	UTR_3_PRIME	ENST00000431235.2:c.*15A>C	-	12/12	-	-	-	
1	SNV	chr17	75494755	A	C	55	44	11	0.2	SEPT9	ENSG00000184640	UTR_3_PRIME	ENST00000427180.1:c.*15A>C	-	10/10	-	-	-	
1	SNV	chr17	75494755	A	C	55	44	11	0.2	SEPT9	ENSG00000184640	UTR_3_PRIME	ENST00000423034.2:c.*15A>C	-	11/11	-	-	-	
1	SNV	chr17	75494755	A	C	55	44	11	0.2	SEPT9	ENSG00000184640	UTR_3_PRIME	ENST00000329047.8:c.*15A>C	-	11/11	-	-	-	
10	SNV	chr17	75494755	A	C	85	66	19	0.224	SEPT9	ENSG00000184640	UTR_3_PRIME	ENST00000541152.2:c.*15A>C	-	10/10	-	-	-	
10	SNV	chr17	75494755	A	C	85	66	19	0.224	SEPT9	ENSG00000184640	UTR_3_PRIME	ENST00000329047.8:c.*15A>C	-	11/11	-	-	-	
10	SNV	chr17	75494755	A	C	85	66	19	0.224	SEPT9	ENSG00000184640	UTR_3_PRIME	ENST00000427177.1:c.*15A>C	-	12/12	YES	-	-	
10	SNV	chr17	75494755	A	C	85	66	19	0.224	SEPT9	ENSG00000184640	UTR_3_PRIME	ENST00000427674.2:c.*15A>C	-	11/11	-	-	-	
10	SNV	chr17	75494755	A	C	85	66	19	0.224	SEPT9	ENSG00000184640	UTR_3_PRIME	ENST00000427180.1:c.*15A>C	-	10/10	-	-	-	
10	SNV	chr17	75494755	A	C	85	66	19	0.224	SEPT9	ENSG00000184640	UTR_3_PRIME	ENST00000431235.2:c.*15A>C	-	12/12	-	-	-	
10	SNV	chr17	75494755	A	C	85	66	19	0.224	SEPT9	ENSG00000184640	UTR_3_PRIME	ENST00000423034.2:c.*15A>C	-	11/11	-	-	-	
10	SNV	chr17	75494755	A	C	85	66	19	0.224	SEPT9	ENSG00000184640	UTR_3_PRIME	ENST00000449803.2:c.*15A>C	-	12/12	-	-	-	
12	SNV	chr17	75494755	A	C	55	39	16	0.286	SEPT9	ENSG00000184640	UTR_3_PRIME	ENST00000427674.2:c.*15A>C	-	11/11	-	-	-	
12	SNV	chr17	75494755	A	C	55	39	16	0.286	SEPT9	ENSG00000184640	UTR_3_PRIME	ENST00000431235.2:c.*15A>C	-	12/12	-	-	-	
12	SNV	chr17	75494755	A	C	55	39	16	0.286	SEPT9	ENSG00000184640	UTR_3_PRIME	ENST00000329047.8:c.*15A>C	-	11/11	-	-	-	
12	SNV	chr17	75494755	A	C	55	39	16	0.286	SEPT9	ENSG00000184640	UTR_3_PRIME	ENST00000423034.2:c.*15A>C	-	11/11	-	-	-	
12	SNV	chr17	75494755	A	C	55	39	16	0.286	SEPT9	ENSG00000184640	UTR_3_PRIME	ENST00000427177.1:c.*15A>C	-	12/12	YES	-	-	
12	SNV	chr17	75494755	A	C	55	39	16	0.286	SEPT9	ENSG00000184640	UTR_3_PRIME	ENST00000449803.2:c.*15A>C	-	12/12	-	-	-	
12	SNV	chr17	75494755	A	C	55	39	16	0.286	SEPT9	ENSG00000184640	UTR_3_PRIME	ENST00000427180.1:c.*15A>C	-	10/10	-	-	-	
12	SNV	chr17	75494755	A	C	55	39	16	0.286	SEPT9	ENSG00000184640	UTR_3_PRIME	ENST00000541152.2:c.*15A>C	-	10/10	-	-	-	
25	SNV	chr17	75471872	G	C	283	143	140	0.495	SEPT9	ENSG00000184640	NON_SYNONYMOUS_CODING	ENST00000427180.1:c.272G>C	ENSP00000415624.1:p.Gly91Ala	1/10	-	-	-	
32	SNV	chr17	75494755	A	C	59	44	15	0.254	SEPT9	ENSG00000184640	UTR_3_PRIME	ENST00000427180.1:c.*15A>C	-	10/10	-	-	-	
32	SNV	chr17	75494755	A	C	59	44	15	0.254	SEPT9	ENSG00000184640	UTR_3_PRIME	ENST00000541152.2:c.*15A>C	-	10/10	-	-	-	
32	SNV	chr17	75494755	A	C	59	44	15	0.254	SEPT9	ENSG00000184640	UTR_3_PRIME	ENST00000449803.2:c.*15A>C	-	12/12	-	-	-	
32	SNV	chr17	75494755	A	C	59	44	15	0.254	SEPT9	ENSG00000184640	UTR_3_PRIME	ENST00000427177.1:c.*15A>C	-	12/12	YES	-	-	
32	SNV	chr17	75494755	A	C	59	44	15	0.254	SEPT9	ENSG00000184640	UTR_3_PRIME	ENST00000427674.2:c.*15A>C	-	11/11	-	-	-	
32	SNV	chr17	75494755	A	C	59	44	15	0.254	SEPT9	ENSG00000184640	UTR_3_PRIME	ENST00000329047.8:c.*15A>C	-	11/11	-	-	-	
32	SNV	chr17	75494755	A	C	59	44	15	0.254	SEPT9	ENSG00000184640	UTR_3_PRIME	ENST00000423034.2:c.*15A>C	-	11/11	-	-	-	
32	SNV	chr17	75494755	A	C	59	44	15	0.254	SEPT9	ENSG00000184640	UTR_3_PRIME	ENST00000431235.2:c.*15A>C	-	12/12	-	-	-	
63	SNV	chr17	75495157	C	T	227	117	110	0.485	SEPT9	ENSG00000184640	UTR_3_PRIME	ENST00000423034.2:c.*417C>T	-	11/11	-	-	-	
63	SNV	chr17	75495157	C	T	227	117	110	0.485	SEPT9	ENSG00000184640	UTR_3_PRIME	ENST00000427177.1:c.*417C>T	-	12/12	YES	-	-	
63	SNV	chr17	75495157	C	T	227	117	110	0.485	SEPT9	ENSG00000184640	UTR_3_PRIME	ENST00000431235.2:c.*417C>T	-	12/12	-	-	-	
63	SNV	chr17	75495157	C	T	227	117	110	0.485	SEPT9	ENSG00000184640	UTR_3_PRIME	ENST00000427674.2:c.*417C>T	-	11/11	-	-	-	
63	SNV	chr17	75495157	C	T	227	117	110	0.485	SEPT9	ENSG00000184640	UTR_3_PRIME	ENST00000449803.2:c.*417C>T	-	12/12	-	-	-	
63	SNV	chr17	75495157	C	T	227	117	110	0.485	SEPT9	ENSG00000184640	UTR_3_PRIME	ENST00000329047.8:c.*417C>T	-	11/11	-	-	-	
63	SNV	chr17	75495157	C	T	227	117	110	0.485	SEPT9	ENSG00000184640	UTR_3_PRIME	ENST00000541152.2:c.*417C>T	-	10/10	-	-	-	
63	SNV	chr17	75495157	C	T	227	117	110	0.485	SEPT9	ENSG00000184640	UTR_3_PRIME	ENST00000427180.1:c.*417C>T	-	10/10	-	-	-	
S63	SNV	chr9	131447370	C	T	348	179	169	0.486	SET	ENSG00000119335	START_GAINED	ENST00000409104.3:c.-77C>T	-	1/8	-	0.0002	rs536265082	
12	INDEL	chr3	47162644	G	-A	190	129	61	0.321	SETD2	ENSG00000181555	FRAME_SHIFT	ENST00000412450.1:c.3350delC	ENSP00000416401.1:p.Ser1117PhefsTer16	3/3	-	-	-	
12	INDEL	chr3	47162644	G	-A	190	129	61	0.321	SETD2	ENSG00000181555	FRAME_SHIFT	ENST00000409792.3:c.3482delC	ENSP00000386759.3:p.Ser1161PhefsTer16	3/21	YES	-	-	
12	INDEL	chr3	47058645	T	+A	392	251	141	0.358	SETD2	ENSG00000181555	FRAME_SHIFT	-	-	21/21	YES	-	-	
64	SNV	chr3	47155429	T	A	61	47	14	0.229	SETD2	ENSG00000181555	NON_SYNONYMOUS_CODING	ENST00000409792.3:c.4652A>T	ENSP00000386759.3:p.Asp1551Val	5/21	YES	-	-	
S18	SNV	chr3	47164370	G	T	430	227	203	0.472	SETD2	ENSG00000181555	NON_SYNONYMOUS_CODING	ENST00000409792.3:c.1756C>A	ENSP00000386759.3:p.His586Asn	3/21	YES	-	-	
S18	SNV	chr3	47164370	G	T	430	227	203	0.472	SETD2	ENSG00000181555	NON_SYNONYMOUS_CODING	ENST00000412450.1:c.1624C>A	ENSP00000416401.1:p.His542Asn	3/3	-	-	-	
S40	SNV	chr3	47164472	C	G	576	282	294	0.51	SETD2	ENSG00000181555	NON_SYNONYMOUS_CODING	ENST00000412450.1:c.1522G>C	ENSP00000416401.1:p.Ala508Pro	3/3	-	-	-	
S40	SNV	chr3	47164472	C	G	576	282	294	0.51	SETD2	ENSG00000181555	NON_SYNONYMOUS_CODING	ENST00000409792.3:c.1654G>C	ENSP00000386759.3:p.Ala552Pro	3/21	YES	-	-	
S51	SNV	chr3	47164922	G	A	696	344	352	0.505	SETD2	ENSG00000181555	NON_SYNONYMOUS_CODING	ENST00000412450.1:c.1072C>T	ENSP00000416401.1:p.Arg358Trp	3/3	-	-	-	
S51	SNV	chr3	47164922	G	A	696	344	352	0.505	SETD2	ENSG00000181555	NON_SYNONYMOUS_CODING	ENST00000409792.3:c.1204C>T	ENSP00000386759.3:p.Arg402Trp	3/21	YES	-	-	
S59	SNV	chr3	47164124	G	C	586	302	284	0.485	SETD2	ENSG00000181555	NON_SYNONYMOUS_CODING	ENST00000412450.1:c.1870C>G	ENSP00000416401.1:p.Pro624Ala	3/3	-	-	-	
S59	SNV	chr3	47164124	G	C	586	302	284	0.485	SETD2	ENSG00000181555	NON_SYNONYMOUS_CODING	ENST00000409792.3:c.2002C>G	ENSP00000386759.3:p.Pro668Ala	3/21	YES	-	-	
1	SNV	chr2	198266468	T	C	175	126	49	0.28	SF3B1	ENSG00000115524	NON_SYNONYMOUS_CODING	ENST00000335508.6:c.2368A>G	ENSP00000335321.5:p.Lys790Glu	16/25	YES	-	-	
37	SNV	chr2	198266713	C	T	54	31	23	0.426	SF3B1	ENSG00000115524	NON_SYNONYMOUS_CODING	ENST00000335508.6:c.2219G>A	ENSP00000335321.5:p.Gly740Glu	15/25	YES	-	COSM1530097,COSM133120	
43	SNV	chr2	198266834	T	C	200	94	106	0.53	SF3B1	ENSG00000115524	NON_SYNONYMOUS_CODING	ENST00000335508.6:c.2098A>G	ENSP00000335321.5:p.Lys700Glu	15/25	YES	0.0002	rs559063155,COSM84677	
59	SNV	chr2	198266834	T	C	39	20	19	0.487	SF3B1	ENSG00000115524	NON_SYNONYMOUS_CODING	ENST00000335508.6:c.2098A>G	ENSP00000335321.5:p.Lys700Glu	15/25	YES	0.0002	rs559063155,COSM84677	
8	SNV	chr11	236327	A	G	170	62	108	0.635	SIRT3	ENSG00000142082	START_LOST	ENST00000525319.1:c.2T>C	ENSP00000435464.1:p.Met1?	1/7	-	0.0002	rs558345508	
8	SNV	chr11	236327	A	G	170	62	108	0.635	SIRT3	ENSG00000142082	START_LOST	ENST00000382743.4:c.2T>C	ENSP00000372191.4:p.Met1?	1/7	YES	0.0002	rs558345508	
8	SNV	chr11	236327	A	G	170	62	108	0.635	SIRT3	ENSG00000142082	START_LOST	ENST00000532956.1:c.2T>C	ENSP00000433077.1:p.Met1?	1/6	-	0.0002	rs558345508	
8	SNV	chr11	236327	A	G	170	62	108	0.635	SIRT3	ENSG00000142082	START_LOST	ENST00000524564.1:c.2T>C	ENSP00000432937.1:p.Met1?	1/6	-	0.0002	rs558345508	
S18	SNV	chr4	25671457	T	C	435	217	218	0.501	SLC34A2	ENSG00000157765	NON_SYNONYMOUS_CODING	ENST00000504570.1:c.821T>C	ENSP00000425501.1:p.Ile274Thr	7/13	-	-	-	
S18	SNV	chr4	25671457	T	C	435	217	218	0.501	SLC34A2	ENSG00000157765	NON_SYNONYMOUS_CODING	ENST00000382051.3:c.824T>C	ENSP00000371483.3:p.Ile275Thr	7/13	YES	-	-	

S18	SNV	chr4	25671457	T	C	435	217	218	0.501	SLC34A2	ENSG00000157765	NON_SYNONYMOUS_CODING	ENST00000503434.1:c.821T>C	ENSP00000423021.1:p.Ile274Thr	7/13	-	-	-
46	SNV	chr12	21051383	G	A	43	21	22	0.512	SLCO1B3	ENSG00000111700	NON_SYNONYMOUS_CODING	ENST00000261196.2:c.1696G>A	ENSP00000261196.2:p.Glu566Lys	13/15	-	-	-
46	SNV	chr12	21051383	G	A	43	21	22	0.512	SLCO1B3	ENSG00000111700	NON_SYNONYMOUS_CODING	ENST00000544370.1:c.1168G>A	ENSP00000443225.1:p.Glu390Lys	8/10	-	-	-
46	SNV	chr12	21051383	G	A	43	21	22	0.512	SLCO1B3	ENSG00000111700	NON_SYNONYMOUS_CODING	ENST00000381545.3:c.1696G>A	ENSP00000370956.3:p.Glu566Lys	14/16	YES	-	-
44	SNV	chr18	48577727	A	G	241	125	116	0.481	SMAD4	ENSG00000141646	NON_SYNONYMOUS_CODING	ENST00000452201.2:c.458A>G	ENSP00000409551.2:p.Asn153Ser	6/9	-	-	-
S58	SNV	chr6	71377737	G	A	384	205	179	0.466	SMAP1	ENSG00000112305	NON_SYNONYMOUS_CODING	ENST00000316999.5:c.11G>A	ENSP00000313382.5:p.Arg4His	1/10	-	-	-
S58	SNV	chr6	71377737	G	A	384	205	179	0.466	SMAP1	ENSG00000112305	NON_SYNONYMOUS_CODING	ENST00000370452.3:c.11G>A	ENSP00000359481.3:p.Arg4His	1/11	-	-	-
S58	SNV	chr6	71377737	G	A	384	205	179	0.466	SMAP1	ENSG00000112305	NON_SYNONYMOUS_CODING	ENST00000422334.2:c.11G>A	ENSP00000398541.2:p.Arg4His	1/5	-	-	-
S58	SNV	chr6	71377737	G	A	384	205	179	0.466	SMAP1	ENSG00000112305	NON_SYNONYMOUS_CODING	ENST00000370455.3:c.11G>A	ENSP00000359484.3:p.Arg4His	1/11	YES	-	-
25	SNV	chr19	11097598	A	C	71	43	28	0.394	SMARCA4	ENSG00000127616	NON_SYNONYMOUS_CODING	ENST00000450717.3:c.778A>C	ENSP00000397783.2:p.Met260Leu	4/33	-	-	-
25	SNV	chr19	11097598	A	C	71	43	28	0.394	SMARCA4	ENSG00000127616	NON_SYNONYMOUS_CODING	ENST00000413806.3:c.778A>C	ENSP00000414727.2:p.Met260Leu	4/33	-	-	-
25	SNV	chr19	11097598	A	C	71	43	28	0.394	SMARCA4	ENSG00000127616	NON_SYNONYMOUS_CODING	ENST00000444061.3:c.778A>C	ENSP00000392837.2:p.Met260Leu	6/35	-	-	-
25	SNV	chr19	11097598	A	C	71	43	28	0.394	SMARCA4	ENSG00000127616	NON_SYNONYMOUS_CODING	ENST00000429416.3:c.778A>C	ENSP00000395654.1:p.Met260Leu	6/36	YES	-	-
25	SNV	chr19	11097598	A	C	71	43	28	0.394	SMARCA4	ENSG00000127616	NON_SYNONYMOUS_CODING	ENST00000541122.2:c.778A>C	ENSP00000445036.2:p.Met260Leu	6/35	-	-	-
25	SNV	chr19	11097598	A	C	71	43	28	0.394	SMARCA4	ENSG00000127616	NON_SYNONYMOUS_CODING	ENST00000344626.4:c.778A>C	ENSP00000343896.4:p.Met260Leu	5/35	-	-	-
25	SNV	chr19	11097598	A	C	71	43	28	0.394	SMARCA4	ENSG00000127616	NON_SYNONYMOUS_CODING	ENST00000358026.2:c.778A>C	ENSP00000350720.2:p.Met260Leu	5/36	-	-	-
47	INDEL	chr4	186544619	C	+GGT	134	87	47	0.341	SORBS2	ENSG00000154556	CODON_CHANGE_PLUS_CODON_INSERTION	-	-	7/15	-	-	-
47	INDEL	chr4	186544619	C	+GGT	134	87	47	0.341	SORBS2	ENSG00000154556	CODON_CHANGE_PLUS_CODON_INSERTION	-	-	14/22	-	-	-
47	INDEL	chr4	186544619	C	+GGT	134	87	47	0.341	SORBS2	ENSG00000154556	CODON_CHANGE_PLUS_CODON_INSERTION	-	-	13/21	-	-	-
47	INDEL	chr4	186544619	C	+GGT	134	87	47	0.341	SORBS2	ENSG00000154556	CODON_CHANGE_PLUS_CODON_INSERTION	-	-	16/24	YES	-	-
S30	SNV	chr4	186587225	T	C	446	250	196	0.44	SORBS2	ENSG00000154556	NON_SYNONYMOUS_CODING	ENST00000451701.1:c.322A>G	ENSP00000404430.1:p.Ser108Gly	6/6	-	0.0002	rs563735895
S51	SNV	chr4	186599689	T	C	566	290	276	0.488	SORBS2	ENSG00000154556	NON_SYNONYMOUS_CODING	ENST00000449407.2:c.101A>G	ENSP00000397262.2:p.Tyr34Cys	6/21	-	-	COSM3946231
S51	SNV	chr4	186599689	T	C	566	290	276	0.488	SORBS2	ENSG00000154556	NON_SYNONYMOUS_CODING	ENST00000457934.1:c.143A>G	ENSP00000405314.1:p.Tyr48Cys	5/6	-	-	COSM3946231
S51	SNV	chr4	186599689	T	C	566	290	276	0.488	SORBS2	ENSG00000154556	NON_SYNONYMOUS_CODING	ENST00000421420.1:c.143A>G	ENSP00000393258.1:p.Tyr48Cys	4/5	-	-	COSM3946231
S51	SNV	chr4	186599689	T	C	566	290	276	0.488	SORBS2	ENSG00000154556	START_GAINED	ENST00000289776.7:c.-158A>G	-	4/21	-	-	COSM3946231
S51	SNV	chr4	186599689	T	C	566	290	276	0.488	SORBS2	ENSG00000154556	NON_SYNONYMOUS_CODING	ENST00000414724.1:c.101A>G	ENSP00000403417.1:p.Tyr34Cys	5/6	-	-	COSM3946231
S51	SNV	chr4	186599689	T	C	566	290	276	0.488	SORBS2	ENSG00000154556	NON_SYNONYMOUS_CODING	ENST00000415274.1:c.101A>G	ENSP00000415392.1:p.Tyr34Cys	5/6	-	-	COSM3946231
S51	SNV	chr4	186599689	T	C	566	290	276	0.488	SORBS2	ENSG00000154556	START_GAINED	ENST00000444771.1:c.-158A>G	-	3/5	-	-	COSM3946231
S51	SNV	chr4	186599689	T	C	566	290	276	0.488	SORBS2	ENSG00000154556	NON_SYNONYMOUS_CODING	ENST00000355634.5:c.143A>G	ENSP00000347852.5:p.Tyr48Cys	7/24	YES	-	COSM3946231
S51	SNV	chr4	186599689	T	C	566	290	276	0.488	SORBS2	ENSG00000154556	NON_SYNONYMOUS_CODING	ENST00000451958.1:c.50A>G	ENSP00000405092.1:p.Tyr17Cys	5/7	-	-	COSM3946231
S51	SNV	chr4	186599689	T	C	566	290	276	0.488	SORBS2	ENSG00000154556	NON_SYNONYMOUS_CODING	ENST00000432655.1:c.143A>G	ENSP00000407006.1:p.Tyr48Cys	6/7	-	-	COSM3946231
S51	SNV	chr4	186599689	T	C	566	290	276	0.488	SORBS2	ENSG00000154556	NON_SYNONYMOUS_CODING	ENST00000435480.1:c.101A>G	ENSP00000415166.1:p.Tyr34Cys	4/5	-	-	COSM3946231
S51	SNV	chr4	186599689	T	C	566	290	276	0.488	SORBS2	ENSG00000154556	NON_SYNONYMOUS_CODING	ENST00000319471.9:c.101A>G	ENSP00000322182.4:p.Tyr34Cys	5/23	-	-	COSM3946231
S51	SNV	chr4	186599689	T	C	566	290	276	0.488	SORBS2	ENSG00000154556	NON_SYNONYMOUS_CODING	ENST00000452351.1:c.101A>G	ENSP00000412447.1:p.Tyr34Cys	4/4	-	-	COSM3946231
S51	SNV	chr4	186599689	T	C	566	290	276	0.488	SORBS2	ENSG00000154556	NON_SYNONYMOUS_CODING	ENST00000439049.1:c.50A>G	ENSP00000416464.1:p.Tyr17Cys	4/6	-	-	COSM3946231
S51	SNV	chr4	186599689	T	C	566	290	276	0.488	SORBS2	ENSG00000154556	NON_SYNONYMOUS_CODING	ENST00000431902.1:c.101A>G	ENSP00000405432.1:p.Tyr34Cys	5/6	-	-	COSM3946231
S51	SNV	chr4	186599689	T	C	566	290	276	0.488	SORBS2	ENSG00000154556	START_GAINED	ENST00000439914.1:c.-158A>G	-	3/6	-	-	COSM3946231
S51	SNV	chr4	186599689	T	C	566	290	276	0.488	SORBS2	ENSG00000154556	NON_SYNONYMOUS_CODING	ENST00000448662.2:c.50A>G	ENSP00000409158.2:p.Tyr17Cys	4/21	-	-	COSM3946231
S51	SNV	chr4	186599689	T	C	566	290	276	0.488	SORBS2	ENSG00000154556	NON_SYNONYMOUS_CODING	ENST00000420158.1:c.143A>G	ENSP00000410233.1:p.Tyr48Cys	6/7	-	-	COSM3946231
S51	SNV	chr4	186599689	T	C	566	290	276	0.488	SORBS2	ENSG00000154556	NON_SYNONYMOUS_CODING	ENST00000425679.1:c.50A>G	ENSP00000415537.1:p.Tyr17Cys	4/6	-	-	COSM3946231
S51	SNV	chr4	186599689	T	C	566	290	276	0.488	SORBS2	ENSG00000154556	NON_SYNONYMOUS_CODING	ENST00000429056.1:c.101A>G	ENSP00000390162.1:p.Tyr34Cys	6/6	-	-	COSM3946231
S51	SNV	chr4	186599689	T	C	566	290	276	0.488	SORBS2	ENSG00000154556	NON_SYNONYMOUS_CODING	ENST00000419063.1:c.143A>G	ENSP00000408504.1:p.Tyr48Cys	6/7	-	-	COSM3946231
S51	SNV	chr4	186599689	T	C	566	290	276	0.488	SORBS2	ENSG00000154556	START_GAINED	ENST00000456596.1:c.-158A>G	-	4/6	-	-	COSM3946231
S51	SNV	chr4	186599689	T	C	566	290	276	0.488	SORBS2	ENSG00000154556	NON_SYNONYMOUS_CODING	ENST00000428330.1:c.50A>G	ENSP00000402328.1:p.Tyr17Cys	5/6	-	-	COSM3946231
S51	SNV	chr4	186599689	T	C	566	290	276	0.488	SORBS2	ENSG00000154556	START_GAINED	ENST00000431808.1:c.-158A>G	-	5/22	-	-	COSM3946231
S56	SNV	chr4	186578636	G	A	790	400	390	0.494	SORBS2	ENSG00000154556	NON_SYNONYMOUS_CODING	ENST00000319471.9:c.467C>T	ENSP00000322182.9:p.Pro156Leu	7/23	-	-	-
S56	SNV	chr4	186578636	G	A	790	400	390	0.494	SORBS2	ENSG00000154556	NON_SYNONYMOUS_CODING	ENST00000444771.1:c.209C>T	ENSP00000410453.1:p.Pro70Leu	5/5	-	-	-
S56	SNV	chr4	186578636	G	A	790	400	390	0.494	SORBS2	ENSG00000154556	NON_SYNONYMOUS_CODING	ENST00000448662.2:c.416C>T	ENSP00000409158.2:p.Pro139Leu	6/21	-	-	-
S56	SNV	chr4	186578636	G	A	790	400	390	0.494	SORBS2	ENSG00000154556	NON_SYNONYMOUS_CODING	ENST00000284776.7:c.209C>T	ENSP00000284776.7:p.Pro70Leu	6/21	-	-	-
S56	SNV	chr4	186578636	G	A	790	400	390	0.494	SORBS2	ENSG00000154556	NON_SYNONYMOUS_CODING	ENST00000430503.1:c.347C>T	ENSP00000405349.1:p.Pro116Leu	7/7	-	-	-
S56	SNV	chr4	186578636	G	A	790	400	390	0.494	SORBS2	ENSG00000154556	NON_SYNONYMOUS_CODING	ENST00000438278.1:c.39C>T	ENSP00000389995.1:p.Pro14Leu	1/9	-	-	-
S56	SNV	chr4	186578636	G	A	790	400	390	0.494	SORBS2	ENSG00000154556	NON_SYNONYMOUS_CODING	ENST00000355634.5:c.509C>T	ENSP00000347852.5:p.Pro170Leu	9/24	YES	-	-
S56	SNV	chr4	186578636	G	A	790	400	390	0.494	SORBS2	ENSG00000154556	NON_SYNONYMOUS_CODING	ENST00000445343.1:c.209C>T	ENSP00000399048.1:p.Pro70Leu	5/6	-	-	-
S56	SNV	chr4	186578636	G	A	790	400	390	0.494	SORBS2	ENSG00000154556	NON_SYNONYMOUS_CODING	ENST00000393528.3:c.347C>T	ENSP00000377162.3:p.Pro116Leu	7/23	-	-	-
S56	SNV	chr4	186578636	G	A	790	400	390	0.494	SORBS2	ENSG00000154556	NON_SYNONYMOUS_CODING	ENST00000449407.2:c.467C>T	ENSP00000397262.2:p.Pro156Leu	8/21	-	-	-
S56	SNV	chr4	186578636	G	A	790	400	390	0.494	SORBS2	ENSG00000154556	NON_SYNONYMOUS_CODING	ENST00000439914.1:c.209C>T	ENSP00000408909.1:p.Pro70Leu	5/6	-	-	-
S56	SNV	chr4	186578636	G	A	790	400	390	0.494	SORBS2	ENSG00000154556	NON_SYNONYMOUS_CODING	ENST00000450341.1:c.209C>T	ENSP00000415680.1:p.Pro70Leu	6/6	-	-	-
S56	SNV	chr4	186578636	G	A	790	400	390	0.494	SORBS2	ENSG00000154556	NON_SYNONYMOUS_CODING	ENST00000431808.1:c.209C>T	ENSP00000411764.1:p.Pro70Leu	7/22	-	-	-
S56	SNV	chr4	186578636	G	A	790	400	390	0.494	SORBS2	ENSG00000154556	NON_SYNONYMOUS_CODING	ENST00000319454.6:c.347C>T	ENSP00000321983.6:p.Pro116Leu	6/20	-	-	-
S56	SNV	chr4	186578636	G	A	790	400	390	0.494	SORBS2	ENSG00000154556	NON_SYNONYMOUS_CODING	ENST00000437304.2:c.746C>T	ENSP00000396008.2:p.Pro249Leu	7/23	-	-	-
1	SNV	chr17	20108350	G	A	466	276	190	0.408	SPECC1	ENSG00000128487	NON_SYNONYMOUS_CODING	ENST00000395530.2:c.745G>A	ENSP00000378901.2:p.Asp249Asn	2/13	-	-	COSM1302499,COSM1520446,COSM215671
1	SNV	chr17	20108350	G	A	466	276	190	0.408	SPECC1	ENSG00000128487	NON_SYNONYMOUS_CODING	ENST00000395527.4:c.988G>A	ENSP00000378898.4:p.Asp330Asn	4/15	-	-	COSM1302499,COSM1520446,COSM215671
1	SNV	chr17	20108350	G	A	466	276	190	0.408	SPECC1	ENSG00000128487	NON_SYNONYMOUS_CODING	ENST00000395529.3:c.988G>A	ENSP00000378900.3:p.Asp330Asn	4/8	-	-	COSM1302499,COSM1520446,COSM215671
1	SNV	chr17	20108350	G	A	466	276	190	0.408	SPECC1	ENSG00000128487	NON_SYNONYMOUS_CODING	ENST00000261503.5:c.988G>A	ENSP00000261503.5:p.Asp330Asn	4/15	YES	-	COSM1302499,COSM1520446,COSM215671
1	SNV	chr17	20108350	G	A	466	276	190	0.408	SPECC1	ENSG00000128487	NON_SYNONYMOUS_CODING	ENST00000395522.2:c.745G>A	ENSP00000378893.2:p.Asp249Asn	2/6	-	-	COSM1302499,COSM1520446,COSM215671
1	SNV	chr17	20108350	G	A	466	276	190	0.408	SPECC1	ENSG00000128487	NON_SYNONYMOUS_CODING	ENST00000395525.3:c.745G>A	ENSP00000378896.3:p.Asp249Asn	2/6	-	-	COSM1302499,COSM1520446,COSM215671
21	SNV	chr17	74732959	G	T	171	89	82	0.48	SRSF2	ENSG00000161547	NON_SYNONYMOUS_CODING	ENST00000508921.3:c.284C>A	ENSP00000441780.2:p.Pro95His	1/3	-	-	COSM211504,COSM146288,COSM211661,COSM146290,COSM469720,COSM211506,COSM211028,COSM211029,COSM211505
21	SNV	chr17	74732959	G	T	171	89	82	0.48	SRSF2	ENSG00000161547	NON_SYNONYMOUS_CODING	ENST00000358156.6:c.284C>A	ENSP00000350877.6:p.Pro95His	1/2	-	-	COSM211504,COSM146288,COSM211661,COSM146290,COSM469720,COSM211506,COSM211028,COSM211029,COSM211505
21	SNV	chr17	74732959	G	T	171	89	82	0.48	SRSF2	ENSG00000161547	NON_SYNONYMOUS_CODING	ENST00000392485.2:c.284C>A	ENSP00000376276.2:p.Pro95His	1/2	YES	-	COSM211504,COSM146288,COSM211661,COSM146290,COSM469720,COSM211506,COSM211028,COSM211029,COSM211505
29	SNV	chr17	74732959	G	T	160	113	47	0.294	SRSF2	ENSG00000161547	NON_SYNONYMOUS_CODING	ENST00000358156.6:c.284C>A	ENSP00000350877.6:p.Pro95His	1/2	-	-	COSM211504,COSM146288,COSM211661,COSM146290,COSM469720,COSM211506,COSM211028,COSM211029,COSM211505
29	SNV	chr17	74732959	G	T	160	113	47	0.294	SRSF2	ENSG00000161547	NON_SYNONYMOUS_CODING	ENST00000508921.3:c.284C>A	ENSP00000441780.2:p.Pro95His	1/3	-	-	COSM211504,COSM146288,COSM211661,COSM146290,COSM469720,COSM211506,COSM211028,COSM211029,COSM211505
29	SNV	chr17	74732959	G	T	160	113	47	0.294	SRSF2	ENSG00000161547	NON_SYNONYMOUS_CODING	ENST00000392485.2:c.284C>A	ENSP00000376276.2:p.Pro95His	1/2	YES	-	COSM211504,COSM146288,COSM211661,COSM146290,COSM469720,COSM211506,COSM211028,COSM211029,COSM211505
36	SNV	chr17	74732959	G	T	187	114	73	0.39	SRSF2	ENSG00000161547	NON_SYNONYMOUS_CODING	ENST00000358156.6:c.284C>A	ENSP00000350877.6:p.Pro95His	1/2	-	-	COSM211504,COSM146288,COSM211661,COSM146290,COSM469720,COSM211506,COSM211028,COSM211029,COSM211505
36	SNV	chr17	74732959	G	T	187	114	73	0.39	SRSF2	ENSG00000161547	NON_SYNONYMOUS_CODING	ENST00000508921.3:c.284C>A	ENSP00000441780.2:p.Pro95His	1/3	-	-	COSM211504,COSM146288,COSM211661,COSM146290,COSM469720,COSM211506,COSM211028,COSM211029,COSM211505
36	SNV	chr17	74732959	G	T	187	114	73	0.39	SRSF2	ENSG00000161547	NON_SYNONYMOUS_CODING	ENST00000392485.2:c.284C>A	ENSP00000376276.2:p.Pro95His	1/2	YES	-	COSM211504,COSM146288,COSM211661,COSM146290,COSM469720,COSM211506,COSM211028,COSM211029,COSM211505
38	SNV	chr17	74732959	G	A	301	166	135	0.448	SRSF2	ENSG00000161547	NON_SYNONYMOUS_CODING	ENST00000358156.6:c.284C>T	ENSP00000350877.6:p.Pro95Leu	1/2	-	-	COSM211504,COSM146288,COSM211661,COSM146290,COSM469720,COSM211506,COSM211028,COSM211029,COSM211505
38	SNV	chr17	74732959	G	A	301	166	135	0.448	SRSF2	ENSG00000161547	NON_SYNONYMOUS_CODING	ENST00000392485.2:c.284C>T	ENSP00000376276.2:p.Pro95Leu	1/2	YES	-	COSM211504,COSM146288,COSM211661,COSM146290,COSM469720,COSM211506,COSM211028,COSM211029,COSM211505
38	SNV	chr17	74732959	G	A	301	166	135	0.448	SRSF2	ENSG00000161547	NON_SYNONYMOUS_CODING	ENST00000508921.3:c.284C>T	ENSP00000441780.2:p.Pro95Leu	1/3	-	-	COSM211504,COSM146288,COSM211661,COSM146290,COSM469720,COSM211506,COSM211028,COSM211029,COSM211505
48	SNV	chr17	74732959	G	T	284	147	137	0.482	SRSF2	ENSG00000161547	NON_SYNONYMOUS_CODING	ENST00000508921.3:c.284C>A	ENSP00000441780.2:p.Pro95His	1/3	-	-	COSM211504,COSM146288,COSM211661,COSM146290,COSM469720,COSM211506,COSM211028,COSM211029,COSM211505
48	SNV	chr17	74732959	G	T	284	147	137	0.482	SRSF2	ENSG00000161547	NON_SYNONYMOUS_CODING	ENST00000392485.2:c.284C>A	ENSP00000376276.2:p.Pro95His	1/2	YES	-	COSM211504,COSM146288,COSM211661,COSM146290,COSM469720,COSM211506,COSM211028,COSM211029,COSM211505
48	SNV	chr17	74732959	G	T	284	147	137	0.482	SRSF2	ENSG00000161547	NON_SYNONYMOUS_CODING	ENST00000358156.6:c.284C>A	ENSP00000350877.6:p.Pro95His	1/2	-	-	COSM211504,COSM146288,COSM211661,COSM146290,COSM469720,COSM211506,COSM211028,COSM211029,COSM211505
6	SNV	chr17	74732959	G	T	343	198	145	0.419	SRSF2	ENSG00000161547	NON_SYNONYMOUS_CODING	ENST00000392485.2:c.284C>A	ENSP00000376276.2:p.Pro95His	1/2	YES	-	COSM211504,COSM146288,COSM211661,COSM146290,COSM469720,COSM211506,COSM211028,COSM211029,COSM211505
6	SNV	chr17	74732959	G	T	343	198	145	0.419	SRSF2	ENSG00000161547	NON_SYNONYMOUS_CODING	ENST00000508921.3:c.284C>A	ENSP00000441780.2:p.Pro95His	1/3	-	-	COSM211504,COSM146288,COSM211661,COSM146290,COSM469720,COSM211506,COSM211028,COSM211029,COSM211505

6	SNV	chr17	74732959	G	T	343	198	145	0.419	SRSF2	ENSG00000161547	NON_SYNONYMOUS_CODING	ENST00000358156.6:c.284C>A	ENSP00000350877.6:p.Pro95His	1/2	-	-	COSM211504,COSM146288,COSM211661,COSM146290,COSM4169720,COSM211506,COSM211028,COSM211029,COSM211505
62	SNV	chr17	74732960	G	C	129	56	73	0.566	SRSF2	ENSG00000161547	NON_SYNONYMOUS_CODING	ENST00000508921.3:c.283C>G	ENSP00000441780.2:p.Pro95Ala	1/3	-	-	COSM307352,COSM307353
62	SNV	chr17	74732960	G	C	129	56	73	0.566	SRSF2	ENSG00000161547	NON_SYNONYMOUS_CODING	ENST00000392485.2:c.283C>G	ENSP00000376276.2:p.Pro95Ala	1/2	YES	-	COSM307352,COSM307353
62	SNV	chr17	74732960	G	C	129	56	73	0.566	SRSF2	ENSG00000161547	NON_SYNONYMOUS_CODING	ENST00000358156.6:c.283C>G	ENSP00000350877.6:p.Pro95Ala	1/2	-	-	COSM307352,COSM307353
63	SNV	chr17	74732959	G	C	282	149	133	0.472	SRSF2	ENSG00000161547	NON_SYNONYMOUS_CODING	ENST00000392485.2:c.284C>G	ENSP00000376276.2:p.Pro95Arg	1/2	YES	-	COSM211504,COSM146288,COSM211661,COSM146290,COSM4169720,COSM211506,COSM211028,COSM211029,COSM211505
63	SNV	chr17	74732959	G	C	282	149	133	0.472	SRSF2	ENSG00000161547	NON_SYNONYMOUS_CODING	ENST00000358156.6:c.284C>G	ENSP00000350877.6:p.Pro95Arg	1/2	-	-	COSM211504,COSM146288,COSM211661,COSM146290,COSM4169720,COSM211506,COSM211028,COSM211029,COSM211505
63	SNV	chr17	74732959	G	C	282	149	133	0.472	SRSF2	ENSG00000161547	NON_SYNONYMOUS_CODING	ENST00000508921.3:c.284C>G	ENSP00000441780.2:p.Pro95Arg	1/3	-	-	COSM211504,COSM146288,COSM211661,COSM146290,COSM4169720,COSM211506,COSM211028,COSM211029,COSM211505
7	SNV	chr17	74732959	G	C	261	144	117	0.447	SRSF2	ENSG00000161547	NON_SYNONYMOUS_CODING	ENST00000508921.3:c.284C>G	ENSP00000441780.2:p.Pro95Arg	1/3	-	-	COSM211504,COSM146288,COSM211661,COSM146290,COSM4169720,COSM211506,COSM211028,COSM211029,COSM211505
7	SNV	chr17	74732959	G	C	261	144	117	0.447	SRSF2	ENSG00000161547	NON_SYNONYMOUS_CODING	ENST00000358156.6:c.284C>G	ENSP00000350877.6:p.Pro95Arg	1/2	-	-	COSM211504,COSM146288,COSM211661,COSM146290,COSM4169720,COSM211506,COSM211028,COSM211029,COSM211505
7	SNV	chr17	74732959	G	C	261	144	117	0.447	SRSF2	ENSG00000161547	NON_SYNONYMOUS_CODING	ENST00000392485.2:c.284C>G	ENSP00000376276.2:p.Pro95Arg	1/2	YES	-	COSM211504,COSM146288,COSM211661,COSM146290,COSM4169720,COSM211506,COSM211028,COSM211029,COSM211505
S12	SNV	chr17	74732959	G	T	226	102	124	0.549	SRSF2	ENSG00000161547	NON_SYNONYMOUS_CODING	ENST00000508921.3:c.284C>A	ENSP00000441780.2:p.Pro95His	1/3	-	-	COSM211504,COSM146288,COSM211661,COSM146290,COSM4169720,COSM211506,COSM211028,COSM211029,COSM211505
S12	SNV	chr17	74732959	G	T	226	102	124	0.549	SRSF2	ENSG00000161547	NON_SYNONYMOUS_CODING	ENST00000358156.6:c.284C>A	ENSP00000350877.6:p.Pro95His	1/2	-	-	COSM211504,COSM146288,COSM211661,COSM146290,COSM4169720,COSM211506,COSM211028,COSM211029,COSM211505
S12	SNV	chr17	74732959	G	T	226	102	124	0.549	SRSF2	ENSG00000161547	NON_SYNONYMOUS_CODING	ENST00000392485.2:c.284C>A	ENSP00000376276.2:p.Pro95His	1/2	YES	-	COSM211504,COSM146288,COSM211661,COSM146290,COSM4169720,COSM211506,COSM211028,COSM211029,COSM211505
S12	SNV	chr17	74732959	G	T	226	102	124	0.549	SRSF2	ENSG00000161547	NON_SYNONYMOUS_CODING	ENST00000583836.1:c.284C>A	ENSP00000463317.1:p.Pro95His	1/2	-	-	COSM211504,COSM146288,COSM211661,COSM146290,COSM4169720,COSM211506,COSM211028,COSM211029,COSM211505
S12	SNV	chr17	74732959	G	T	226	102	124	0.549	SRSF2	ENSG00000161547	NON_SYNONYMOUS_CODING	ENST00000359995.5:c.284C>A	ENSP00000353089.5:p.Pro95His	1/3	-	-	COSM211504,COSM146288,COSM211661,COSM146290,COSM4169720,COSM211506,COSM211028,COSM211029,COSM211505
S14	SNV	chr17	74732959	G	C	158	85	73	0.459	SRSF2	ENSG00000161547	NON_SYNONYMOUS_CODING	ENST00000359995.5:c.284C>G	ENSP00000353089.5:p.Pro95Arg	1/3	-	-	COSM211504,COSM146288,COSM211661,COSM146290,COSM4169720,COSM211506,COSM211028,COSM211029,COSM211505
S14	SNV	chr17	74732959	G	C	158	85	73	0.459	SRSF2	ENSG00000161547	NON_SYNONYMOUS_CODING	ENST00000358156.6:c.284C>G	ENSP00000350877.6:p.Pro95Arg	1/2	-	-	COSM211504,COSM146288,COSM211661,COSM146290,COSM4169720,COSM211506,COSM211028,COSM211029,COSM211505
S14	SNV	chr17	74732959	G	C	158	85	73	0.459	SRSF2	ENSG00000161547	NON_SYNONYMOUS_CODING	ENST00000392485.2:c.284C>G	ENSP00000376276.2:p.Pro95Arg	1/2	YES	-	COSM211504,COSM146288,COSM211661,COSM146290,COSM4169720,COSM211506,COSM211028,COSM211029,COSM211505
S14	SNV	chr17	74732959	G	C	158	85	73	0.459	SRSF2	ENSG00000161547	NON_SYNONYMOUS_CODING	ENST00000583836.1:c.284C>G	ENSP00000463317.1:p.Pro95Arg	1/2	-	-	COSM211504,COSM146288,COSM211661,COSM146290,COSM4169720,COSM211506,COSM211028,COSM211029,COSM211505
S14	SNV	chr17	74732959	G	C	158	85	73	0.459	SRSF2	ENSG00000161547	NON_SYNONYMOUS_CODING	ENST00000508921.3:c.284C>G	ENSP00000441780.2:p.Pro95Arg	1/3	-	-	COSM211504,COSM146288,COSM211661,COSM146290,COSM4169720,COSM211506,COSM211028,COSM211029,COSM211505
S18	SNV	chr17	74732959	G	T	294	148	146	0.497	SRSF2	ENSG00000161547	NON_SYNONYMOUS_CODING	ENST00000358156.6:c.284C>A	ENSP00000350877.6:p.Pro95His	1/2	-	-	COSM211504,COSM146288,COSM211661,COSM146290,COSM4169720,COSM211506,COSM211028,COSM211029,COSM211505
S18	SNV	chr17	74732959	G	T	294	148	146	0.497	SRSF2	ENSG00000161547	NON_SYNONYMOUS_CODING	ENST00000508921.3:c.284C>A	ENSP00000441780.2:p.Pro95His	1/3	-	-	COSM211504,COSM146288,COSM211661,COSM146290,COSM4169720,COSM211506,COSM211028,COSM211029,COSM211505
S18	SNV	chr17	74732959	G	T	294	148	146	0.497	SRSF2	ENSG00000161547	NON_SYNONYMOUS_CODING	ENST00000392485.2:c.284C>A	ENSP00000376276.2:p.Pro95His	1/2	YES	-	COSM211504,COSM146288,COSM211661,COSM146290,COSM4169720,COSM211506,COSM211028,COSM211029,COSM211505
S18	SNV	chr17	74732959	G	T	294	148	146	0.497	SRSF2	ENSG00000161547	NON_SYNONYMOUS_CODING	ENST00000583836.1:c.284C>A	ENSP00000463317.1:p.Pro95His	1/2	-	-	COSM211504,COSM146288,COSM211661,COSM146290,COSM4169720,COSM211506,COSM211028,COSM211029,COSM211505
S18	SNV	chr17	74732959	G	T	294	148	146	0.497	SRSF2	ENSG00000161547	NON_SYNONYMOUS_CODING	ENST00000359995.5:c.284C>A	ENSP00000353089.5:p.Pro95His	1/3	-	-	COSM211504,COSM146288,COSM211661,COSM146290,COSM4169720,COSM211506,COSM211028,COSM211029,COSM211505
S19	SNV	chr17	74732959	G	A	361	209	152	0.421	SRSF2	ENSG00000161547	NON_SYNONYMOUS_CODING	ENST00000358156.6:c.284C>T	ENSP00000350877.6:p.Pro95Leu	1/2	-	-	COSM211504,COSM146288,COSM211661,COSM146290,COSM4169720,COSM211506,COSM211028,COSM211029,COSM211505
S19	SNV	chr17	74732959	G	A	361	209	152	0.421	SRSF2	ENSG00000161547	NON_SYNONYMOUS_CODING	ENST00000359995.5:c.284C>T	ENSP00000353089.5:p.Pro95Leu	1/3	-	-	COSM211504,COSM146288,COSM211661,COSM146290,COSM4169720,COSM211506,COSM211028,COSM211029,COSM211505
S19	SNV	chr17	74732959	G	A	361	209	152	0.421	SRSF2	ENSG00000161547	NON_SYNONYMOUS_CODING	ENST00000508921.3:c.284C>T	ENSP00000441780.2:p.Pro95Leu	1/3	-	-	COSM211504,COSM146288,COSM211661,COSM146290,COSM4169720,COSM211506,COSM211028,COSM211029,COSM211505
S19	SNV	chr17	74732959	G	A	361	209	152	0.421	SRSF2	ENSG00000161547	NON_SYNONYMOUS_CODING	ENST00000583836.1:c.284C>T	ENSP00000463317.1:p.Pro95Leu	1/2	-	-	COSM211504,COSM146288,COSM211661,COSM146290,COSM4169720,COSM211506,COSM211028,COSM211029,COSM211505
S19	SNV	chr17	74732959	G	A	361	209	152	0.421	SRSF2	ENSG00000161547	NON_SYNONYMOUS_CODING	ENST00000392485.2:c.284C>T	ENSP00000376276.2:p.Pro95Leu	1/2	YES	-	COSM211504,COSM146288,COSM211661,COSM146290,COSM4169720,COSM211506,COSM211028,COSM211029,COSM211505
S34	SNV	chr17	74732959	G	C	404	262	142	0.352	SRSF2	ENSG00000161547	NON_SYNONYMOUS_CODING	ENST00000508921.3:c.284C>G	ENSP00000441780.2:p.Pro95Arg	1/3	-	-	COSM211504,COSM146288,COSM211661,COSM146290,COSM4169720,COSM211506,COSM211028,COSM211029,COSM211505
S34	SNV	chr17	74732959	G	C	404	262	142	0.352	SRSF2	ENSG00000161547	NON_SYNONYMOUS_CODING	ENST00000359995.5:c.284C>G	ENSP00000353089.5:p.Pro95Arg	1/3	-	-	COSM211504,COSM146288,COSM211661,COSM146290,COSM4169720,COSM211506,COSM211028,COSM211029,COSM211505
S34	SNV	chr17	74732959	G	C	404	262	142	0.352	SRSF2	ENSG00000161547	NON_SYNONYMOUS_CODING	ENST00000583836.1:c.284C>G	ENSP00000463317.1:p.Pro95Arg	1/2	-	-	COSM211504,COSM146288,COSM211661,COSM146290,COSM4169720,COSM211506,COSM211028,COSM211029,COSM211505
S34	SNV	chr17	74732959	G	C	404	262	142	0.352	SRSF2	ENSG00000161547	NON_SYNONYMOUS_CODING	ENST00000358156.6:c.284C>G	ENSP00000350877.6:p.Pro95Arg	1/2	-	-	COSM211504,COSM146288,COSM211661,COSM146290,COSM4169720,COSM211506,COSM211028,COSM211029,COSM211505
S34	SNV	chr17	74732959	G	C	404	262	142	0.352	SRSF2	ENSG00000161547	NON_SYNONYMOUS_CODING	ENST00000392485.2:c.284C>G	ENSP00000376276.2:p.Pro95Arg	1/2	YES	-	COSM211504,COSM146288,COSM211661,COSM146290,COSM4169720,COSM211506,COSM211028,COSM211029,COSM211505
S35	SNV	chr17	74732960	G	T	306	154	152	0.497	SRSF2	ENSG00000161547	NON_SYNONYMOUS_CODING	ENST00000508921.3:c.283C>A	ENSP00000441780.2:p.Pro95Thr	1/3	-	-	COSM307352,COSM307353
S35	SNV	chr17	74732960	G	T	306	154	152	0.497	SRSF2	ENSG00000161547	NON_SYNONYMOUS_CODING	ENST00000583836.1:c.283C>A	ENSP00000463317.1:p.Pro95Thr	1/2	-	-	COSM307352,COSM307353
S35	SNV	chr17	74732960	G	T	306	154	152	0.497	SRSF2	ENSG00000161547	NON_SYNONYMOUS_CODING	ENST00000359995.5:c.283C>A	ENSP00000353089.5:p.Pro95Thr	1/3	-	-	COSM307352,COSM307353
S35	SNV	chr17	74732960	G	T	306	154	152	0.497	SRSF2	ENSG00000161547	NON_SYNONYMOUS_CODING	ENST00000392485.2:c.283C>A	ENSP00000376276.2:p.Pro95Thr	1/2	YES	-	COSM307352,COSM307353
S35	SNV	chr17	74732960	G	T	306	154	152	0.497	SRSF2	ENSG00000161547	NON_SYNONYMOUS_CODING	ENST00000358156.6:c.283C>A	ENSP00000350877.6:p.Pro95Thr	1/2	-	-	COSM307352,COSM307353
S43	INDEL	chr17	74732935	C	- GGCGGCTGTGGTGT GAGTCCGGGGG	258	200	58	0.225	SRSF2	ENSG00000161547	CODON_CHANGE_PLUS_CODON_DELETION	ENST00000583836.1:c.311delG	ENSP00000463317.1:p.Gly104AspfsTer?	1/2	-	-	-
S43	INDEL	chr17	74732935	C	- GGCGGCTGTGGTGT GAGTCCGGGG	258	200	58	0.225	SRSF2	ENSG00000161547	CODON_CHANGE_PLUS_CODON_DELETION	ENST00000392485.2:c.311delG	ENSP00000376276.2:p.Gly104AspfsTer140	1/2	YES	-	-
S43	INDEL	chr17	74732935	C	- GGCGGCTGTGGTGT GAGTCCGGGG	258	200	58	0.225	SRSF2	ENSG00000161547	CODON_CHANGE_PLUS_CODON_DELETION	ENST00000358156.6:c.311delG	ENSP00000350877.6:p.Gly104AspfsTer?	1/2	-	-	-
S43	INDEL	chr17	74732935	C	- GGCGGCTGTGGTGT GAGTCCGGGG	258	200	58	0.225	SRSF2	ENSG00000161547	CODON_CHANGE_PLUS_CODON_DELETION	ENST00000508921.3:c.311delG	ENSP00000441780.2:p.Gly104AspfsTer116	1/3	-	-	-
S43	INDEL	chr17	74732935	C	- GGCGGCTGTGGTGT GAGTCCGGGG	258	200	58	0.225	SRSF2	ENSG00000161547	CODON_CHANGE_PLUS_CODON_DELETION	ENST00000359995.5:c.311delG	ENSP00000353089.5:p.Gly104AspfsTer128	1/3	-	-	-
S51	SNV	chr17	74732959	G	T	334	172	162	0.485	SRSF2	ENSG00000161547	NON_SYNONYMOUS_CODING	ENST00000359995.5:c.284C>A	ENSP00000353089.5:p.Pro95His	1/3	-	-	COSM211504,COSM146288,COSM211661,COSM146290,COSM4169720,COSM211506,COSM211028,COSM211029,COSM211505
S51	SNV	chr17	74732959	G	T	334	172	162	0.485	SRSF2	ENSG00000161547	NON_SYNONYMOUS_CODING	ENST00000583836.1:c.284C>A	ENSP00000463317.1:p.Pro95His	1/2	-	-	COSM211504,COSM146288,COSM211661,COSM146290,COSM4169720,COSM211506,COSM211028,COSM211029,COSM211505
S51	SNV	chr17	74732959	G	T	334	172	162	0.485	SRSF2	ENSG00000161547	NON_SYNONYMOUS_CODING	ENST00000508921.3:c.284C>A	ENSP00000441780.2:p.Pro95His	1/3	-	-	COSM211504,COSM146288,COSM211661,COSM146290,COSM4169720,COSM211506,COSM211028,COSM211029,COSM211505
S51	SNV	chr17	74732959	G	T	334	172	162	0.485	SRSF2	ENSG00000161547	NON_SYNONYMOUS_CODING	ENST00000392485.2:c.284C>A	ENSP00000376276.2:p.Pro95His	1/2	YES	-	COSM211504,COSM146288,COSM211661,COSM146290,COSM4169720,COSM211506,COSM211028,COSM211029,COSM211505
S51	SNV	chr17	74732959	G	T	334	172	162	0.485	SRSF2	ENSG00000161547	NON_SYNONYMOUS_CODING	ENST00000358156.6:c.284C>A	ENSP00000350877.6:p.Pro95His	1/2	-	-	COSM211504,COSM146288,COSM211661,COSM146290,COSM4169720,COSM211506,COSM211028,COSM211029,COSM211505
S53	SNV	chr17	74732959	G	C	374	218	156	0.417	SRSF2	ENSG00000161547	NON_SYNONYMOUS_CODING	ENST00000392485.2:c.284C>G	ENSP00000376276.2:p.Pro95Arg	1/2	YES	-	COSM211504,COSM146288,COSM211661,COSM146290,COSM4169720,COSM211506,COSM211028,COSM211029,COSM211505
S53	SNV	chr17	74732959	G	C	374	218	156	0.417	SRSF2	ENSG00000161547	NON_SYNONYMOUS_CODING	ENST00000508921.3:c.284C>G	ENSP00000441780.2:p.Pro95Arg	1/3	-	-	COSM211504,COSM146288,COSM211661,COSM146290,COSM4169720,COSM211506,COSM211028,COSM211029,COSM211505
S53	SNV	chr17	74732959	G	C	374	218	156	0.417	SRSF2	ENSG00000161547	NON_SYNONYMOUS_CODING	ENST00000359995.5:c.284C>G	ENSP00000353089.5:p.Pro95Arg	1/3	-	-	COSM211504,COSM146288,COSM211661,COSM146290,COSM4169720,COSM211506,COSM211028,COSM211029,COSM211505
S53	SNV	chr17	74732959	G	C	374	218	156	0.417	SRSF2	ENSG00000161547	NON_SYNONYMOUS_CODING	ENST00000358156.6:c.284C>G	ENSP00000350877.6:p.Pro95Arg	1/2	-	-	COSM211504,COSM146288,COSM211661,COSM146290,COSM4169720,COSM211506,COSM211028,COSM211029,COSM211505

S53	SNV	chr17	74732959	G	C	374	218	156	0.417	SRSF2	ENSG00000161547	NON_SYNONYMOUS_CODING	ENST00000583836.1:c.284C>G	ENSP00000463317.1:p.Pro95Arg	1/2	-	-	COSM211504,COSM146288,COSM211661,COSM146290,COSM4169720,COSM211506,COSM211028,COSM211029,COSM211505
S56	SNV	chr17	74732959	G	T	657	365	292	0.444	SRSF2	ENSG00000161547	NON_SYNONYMOUS_CODING	ENST00000583836.1:c.284C>A	ENSP00000463317.1:p.Pro95His	1/2	-	-	COSM211504,COSM146288,COSM211661,COSM146290,COSM4169720,COSM211506,COSM211028,COSM211029,COSM211505
S56	SNV	chr17	74732959	G	T	657	365	292	0.444	SRSF2	ENSG00000161547	NON_SYNONYMOUS_CODING	ENST00000508921.3:c.284C>A	ENSP00000441780.2:p.Pro95His	1/3	-	-	COSM211504,COSM146288,COSM211661,COSM146290,COSM4169720,COSM211506,COSM211028,COSM211029,COSM211505
S56	SNV	chr17	74732959	G	T	657	365	292	0.444	SRSF2	ENSG00000161547	NON_SYNONYMOUS_CODING	ENST00000358156.6:c.284C>A	ENSP00000350877.6:p.Pro95His	1/2	-	-	COSM211504,COSM146288,COSM211661,COSM146290,COSM4169720,COSM211506,COSM211028,COSM211029,COSM211505
S56	SNV	chr17	74732959	G	T	657	365	292	0.444	SRSF2	ENSG00000161547	NON_SYNONYMOUS_CODING	ENST00000392485.2:c.284C>A	ENSP00000376276.2:p.Pro95His	1/2	YES	-	COSM211504,COSM146288,COSM211661,COSM146290,COSM4169720,COSM211506,COSM211028,COSM211029,COSM211505
S56	SNV	chr17	74732959	G	T	657	365	292	0.444	SRSF2	ENSG00000161547	NON_SYNONYMOUS_CODING	ENST00000359995.5:c.284C>A	ENSP00000353089.5:p.Pro95His	1/3	-	-	COSM211504,COSM146288,COSM211661,COSM146290,COSM4169720,COSM211506,COSM211028,COSM211029,COSM211505
S61	SNV	chr17	74732959	G	T	541	283	258	0.477	SRSF2	ENSG00000161547	NON_SYNONYMOUS_CODING	ENST00000508921.3:c.284C>A	ENSP00000441780.2:p.Pro95His	1/3	-	-	COSM211504,COSM146288,COSM211661,COSM146290,COSM4169720,COSM211506,COSM211028,COSM211029,COSM211505
S61	SNV	chr17	74732959	G	T	541	283	258	0.477	SRSF2	ENSG00000161547	NON_SYNONYMOUS_CODING	ENST00000359995.5:c.284C>A	ENSP00000353089.5:p.Pro95His	1/3	-	-	COSM211504,COSM146288,COSM211661,COSM146290,COSM4169720,COSM211506,COSM211028,COSM211029,COSM211505
S61	SNV	chr17	74732959	G	T	541	283	258	0.477	SRSF2	ENSG00000161547	NON_SYNONYMOUS_CODING	ENST00000583836.1:c.284C>A	ENSP00000463317.1:p.Pro95His	1/2	-	-	COSM211504,COSM146288,COSM211661,COSM146290,COSM4169720,COSM211506,COSM211028,COSM211029,COSM211505
S61	SNV	chr17	74732959	G	T	541	283	258	0.477	SRSF2	ENSG00000161547	NON_SYNONYMOUS_CODING	ENST00000392485.2:c.284C>A	ENSP00000376276.2:p.Pro95His	1/2	YES	-	COSM211504,COSM146288,COSM211661,COSM146290,COSM4169720,COSM211506,COSM211028,COSM211029,COSM211505
S61	SNV	chr17	74732959	G	T	541	283	258	0.477	SRSF2	ENSG00000161547	NON_SYNONYMOUS_CODING	ENST00000358156.6:c.284C>A	ENSP00000350877.6:p.Pro95His	1/2	-	-	COSM211504,COSM146288,COSM211661,COSM146290,COSM4169720,COSM211506,COSM211028,COSM211029,COSM211505
S64	SNV	chr17	74732959	G	A	512	274	238	0.465	SRSF2	ENSG00000161547	NON_SYNONYMOUS_CODING	ENST00000359995.5:c.284C>T	ENSP00000353089.5:p.Pro95Leu	1/3	-	-	COSM211504,COSM146288,COSM211661,COSM146290,COSM4169720,COSM211506,COSM211028,COSM211029,COSM211505
S64	SNV	chr17	74732959	G	A	512	274	238	0.465	SRSF2	ENSG00000161547	NON_SYNONYMOUS_CODING	ENST00000583836.1:c.284C>T	ENSP00000463317.1:p.Pro95Leu	1/2	-	-	COSM211504,COSM146288,COSM211661,COSM146290,COSM4169720,COSM211506,COSM211028,COSM211029,COSM211505
S64	SNV	chr17	74732959	G	A	512	274	238	0.465	SRSF2	ENSG00000161547	NON_SYNONYMOUS_CODING	ENST00000508921.3:c.284C>T	ENSP00000441780.2:p.Pro95Leu	1/3	-	-	COSM211504,COSM146288,COSM211661,COSM146290,COSM4169720,COSM211506,COSM211028,COSM211029,COSM211505
S64	SNV	chr17	74732959	G	A	512	274	238	0.465	SRSF2	ENSG00000161547	NON_SYNONYMOUS_CODING	ENST00000358156.6:c.284C>T	ENSP00000350877.6:p.Pro95Leu	1/2	-	-	COSM211504,COSM146288,COSM211661,COSM146290,COSM4169720,COSM211506,COSM211028,COSM211029,COSM211505
S64	SNV	chr17	74732959	G	A	512	274	238	0.465	SRSF2	ENSG00000161547	NON_SYNONYMOUS_CODING	ENST00000392485.2:c.284C>T	ENSP00000376276.2:p.Pro95Leu	1/2	YES	-	COSM211504,COSM146288,COSM211661,COSM146290,COSM4169720,COSM211506,COSM211028,COSM211029,COSM211505
1	SNV	chr18	23618535	C	G	253	144	109	0.431	SS18	ENSG00000141380	NON_SYNONYMOUS_CODING	ENST00000415083.2:c.864G>C	ENSP00000414516.2:p.Gln288His	7/11	YES	0.0020	rs547058660
1	SNV	chr18	23618535	C	G	253	144	109	0.431	SS18	ENSG00000141380	NON_SYNONYMOUS_CODING	ENST00000542420.2:c.795G>C	ENSP00000438066.2:p.Gln265His	7/11	-	0.0020	rs547058660
1	SNV	chr18	23618535	C	G	253	144	109	0.431	SS18	ENSG00000141380	NON_SYNONYMOUS_CODING	ENST00000542743.1:c.708G>C	ENSP00000444551.1:p.Gln236His	8/11	-	0.0020	rs547058660
1	SNV	chr18	23618535	C	G	253	144	109	0.431	SS18	ENSG00000141380	NON_SYNONYMOUS_CODING	ENST00000543952.1:c.708G>C	ENSP00000443097.1:p.Gln236His	8/11	-	0.0020	rs547058660
1	SNV	chr18	23618535	C	G	253	144	109	0.431	SS18	ENSG00000141380	NON_SYNONYMOUS_CODING	ENST00000539849.1:c.618G>C	ENSP00000444647.1:p.Gln206His	6/10	-	0.0020	rs547058660
1	SNV	chr18	23618535	C	G	253	144	109	0.431	SS18	ENSG00000141380	NON_SYNONYMOUS_CODING	ENST00000269137.7:c.864G>C	ENSP00000269137.7:p.Gln288His	7/10	-	0.0020	rs547058660
37	SNV	chr15	57565283	G	T	121	81	40	0.328	TCF12	ENSG00000140262	STOP_GAINED	ENST00000343827.3:c.1219G>T	ENSP00000342459.3:p.Glu407Ter	11/13	-	-	-
37	SNV	chr15	57565283	G	T	121	81	40	0.328	TCF12	ENSG00000140262	STOP_GAINED	ENST00000543579.1:c.1291G>T	ENSP00000440017.1:p.Glu431Ter	12/14	-	-	-
37	SNV	chr15	57565283	G	T	121	81	40	0.328	TCF12	ENSG00000140262	STOP_GAINED	ENST00000267811.5:c.1729G>T	ENSP00000267811.5:p.Glu577Ter	18/20	-	-	-
37	SNV	chr15	57565283	G	T	121	81	40	0.328	TCF12	ENSG00000140262	STOP_GAINED	ENST00000438423.2:c.1801G>T	ENSP00000388940.2:p.Glu601Ter	19/21	YES	-	-
37	SNV	chr15	57565283	G	T	121	81	40	0.328	TCF12	ENSG00000140262	STOP_GAINED	ENST00000452095.2:c.1789G>T	ENSP00000396881.2:p.Glu597Ter	19/21	-	-	-
37	SNV	chr15	57565283	G	T	121	81	40	0.328	TCF12	ENSG00000140262	STOP_GAINED	ENST00000333725.5:c.1801G>T	ENSP00000331057.5:p.Glu601Ter	19/21	-	-	-
37	SNV	chr15	57565283	G	T	121	81	40	0.328	TCF12	ENSG00000140262	STOP_GAINED	ENST00000537840.1:c.1021G>T	ENSP00000444696.1:p.Glu341Ter	10/12	-	-	-
12	SNV	chr19	1650254	G	A	159	87	72	0.453	TCF3	ENSG00000071564	UTR_5_PRIME	ENST00000395423.3:c.-7C>T	-	1/15	-	-	-
12	SNV	chr19	1650254	G	A	159	87	72	0.453	TCF3	ENSG00000071564	UTR_5_PRIME	ENST00000262965.5:c.-7C>T	-	2/19	YES	-	-
12	SNV	chr19	1650254	G	A	159	87	72	0.453	TCF3	ENSG00000071564	UTR_5_PRIME	ENST00000344749.5:c.-7C>T	-	2/19	-	-	-
S06	SNV	chr19	1615284	C	T	90	51	39	0.433	TCF3	ENSG00000071564	NON_SYNONYMOUS_CODING	ENST00000592628.1:c.397G>A	ENSP00000465549.1:p.Glu133Lys	3/5	-	-	-
S06	SNV	chr19	1615284	C	T	90	51	39	0.433	TCF3	ENSG00000071564	SPLICE_SITE_REGION	ENST00000262965.5:c.1822G>A	ENSP00000262965.4:p.Glu608Lys	18/19	YES	-	-
S06	SNV	chr19	1615284	C	T	90	51	39	0.433	TCF3	ENSG00000071564	SPLICE_SITE_REGION	ENST00000395423.3:c.1834G>A	ENSP00000378813.3:p.Glu612Lys	14/15	-	-	-
S06	SNV	chr19	1615284	C	T	90	51	39	0.433	TCF3	ENSG00000071564	NON_SYNONYMOUS_CODING	ENST00000593064.1:c.471G>A	ENSP00000468481.1:p.Glu158Lys	3/5	-	-	-
S06	SNV	chr19	1615284	C	T	90	51	39	0.433	TCF3	ENSG00000071564	NON_SYNONYMOUS_CODING	ENST00000587425.1:c.349G>A	ENSP00000467508.1:p.Glu117Lys	2/4	-	-	-
S06	SNV	chr19	1615284	C	T	90	51	39	0.433	TCF3	ENSG00000071564	SPLICE_SITE_REGION	ENST00000587425.1:c.349G>A	ENSP00000467508.1:p.Glu117Lys	2/4	-	-	-
S06	SNV	chr19	1615284	C	T	90	51	39	0.433	TCF3	ENSG00000071564	SPLICE_SITE_REGION	ENST00000592628.1:c.397G>A	ENSP00000465549.1:p.Glu133Lys	3/5	-	-	-
S06	SNV	chr19	1615284	C	T	90	51	39	0.433	TCF3	ENSG00000071564	SPLICE_SITE_REGION	ENST00000593064.1:c.471G>A	ENSP00000468481.1:p.Glu158Lys	3/5	-	-	-
S06	SNV	chr19	1615284	C	T	90	51	39	0.433	TCF3	ENSG00000071564	NON_SYNONYMOUS_CODING	ENST00000262965.5:c.1822G>A	ENSP00000262965.4:p.Glu608Lys	18/19	YES	-	-
S06	SNV	chr19	1615284	C	T	90	51	39	0.433	TCF3	ENSG00000071564	NON_SYNONYMOUS_CODING	ENST00000395423.3:c.1834G>A	ENSP00000378813.3:p.Glu612Lys	14/15	-	-	-
S37	SNV	chr19	1619835	G	C	128	72	56	0.438	TCF3	ENSG00000071564	NON_SYNONYMOUS_CODING	ENST00000395423.3:c.958C>G	ENSP00000378813.3:p.Arg320Gly	11/15	-	-	COSM1391195,COSM1391196
S37	SNV	chr19	1619835	G	C	128	72	56	0.438	TCF3	ENSG00000071564	NON_SYNONYMOUS_CODING	ENST00000586410.1:c.98C>G	ENSP00000467388.1:p.Arg34Gly	2/3	-	-	COSM1391195,COSM1391196
S37	SNV	chr19	1619835	G	C	128	72	56	0.438	TCF3	ENSG00000071564	NON_SYNONYMOUS_CODING	ENST00000344749.5:c.1111C>G	ENSP00000344375.5:p.Arg371Gly	14/19	-	-	COSM1391195,COSM1391196
S37	SNV	chr19	1619835	G	C	128	72	56	0.438	TCF3	ENSG00000071564	NON_SYNONYMOUS_CODING	ENST00000453954.2:c.859C>G	ENSP00000396363.2:p.Arg287Gly	10/16	-	-	COSM1391195,COSM1391196
S37	SNV	chr19	1619835	G	C	128	72	56	0.438	TCF3	ENSG00000071564	NON_SYNONYMOUS_CODING	ENST00000588136.1:c.1111C>G	ENSP00000468487.1:p.Arg371Gly	13/19	-	-	COSM1391195,COSM1391196
S37	SNV	chr19	1619835	G	C	128	72	56	0.438	TCF3	ENSG00000071564	NON_SYNONYMOUS_CODING	ENST00000262965.5:c.1111C>G	ENSP00000262965.4:p.Arg371Gly	14/19	YES	-	COSM1391195,COSM1391196
1	SNV	chr5	133482299	T	C	90	69	21	0.233	TCF7	ENSG00000081059	UTR_3_PRIME	ENST00000378560.4:c.*4047>C	-	10/10	-	-	-
1	SNV	chr5	133482299	T	C	90	69	21	0.233	TCF7	ENSG00000081059	UTR_3_PRIME	ENST00000395023.1:c.*305T>C	-	9/9	-	-	-
1	SNV	chr5	133482299	T	C	90	69	21	0.233	TCF7	ENSG00000081059	UTR_3_PRIME	ENST00000395029.1:c.*87T>C	-	11/11	-	-	-
1	SNV	chr5	133482299	T	C	90	69	21	0.233	TCF7	ENSG00000081059	UTR_3_PRIME	ENST00000342854.5:c.*305T>C	-	10/10	YES	-	-
S38	SNV	chr5	133478545	G	A	545	357	188	0.345	TCF7	ENSG00000081059	NON_SYNONYMOUS_CODING	ENST00000378564.1:c.889G>A	ENSP00000367827.1:p.Ala297Thr	7/10	-	-	-
S38	SNV	chr5	133478545	G	A	545	357	188	0.345	TCF7	ENSG00000081059	NON_SYNONYMOUS_CODING	ENST00000520699.1:c.63G>A	ENSP00000429935.1:p.Ala22Thr	1/4	-	-	-
S38	SNV	chr5	133478545	G	A	545	357	188	0.345	TCF7	ENSG00000081059	NON_SYNONYMOUS_CODING	ENST00000432532.2:c.544G>A	ENSP00000397946.2:p.Ala182Thr	6/10	-	-	-
S38	SNV	chr5	133478545	G	A	545	357	188	0.345	TCF7	ENSG00000081059	NON_SYNONYMOUS_CODING	ENST00000321603.6:c.889G>A	ENSP00000326654.6:p.Ala297Thr	7/10	-	-	-
S38	SNV	chr5	133478545	G	A	545	357	188	0.345	TCF7	ENSG00000081059	NON_SYNONYMOUS_CODING	ENST00000395029.1:c.889G>A	ENSP00000378472.1:p.Ala297Thr	7/11	-	-	-
S38	SNV	chr5	133478545	G	A	545	357	188	0.345	TCF7	ENSG00000081059	NON_SYNONYMOUS_CODING	ENST00000518915.1:c.544G>A	ENSP00000430179.1:p.Ala182Thr	6/9	-	-	-
S38	SNV	chr5	133478545	G	A	545	357	188	0.345	TCF7	ENSG00000081059	NON_SYNONYMOUS_CODING	ENST00000395023.1:c.544G>A	ENSP00000378469.1:p.Ala182Thr	6/9	-	-	-
S38	SNV	chr5	133478545	G	A	545	357	188	0.345	TCF7	ENSG00000081059	NON_SYNONYMOUS_CODING	ENST00000378560.4:c.544G>A	ENSP00000367822.4:p.Ala182Thr	6/10	-	-	-
S38	SNV	chr5	133478545	G	A	545	357	188	0.345	TCF7	ENSG00000081059	NON_SYNONYMOUS_CODING	ENST00000342854.5:c.889G>A	ENSP00000340347.5:p.Ala297Thr	7/10	YES	-	-
S38	SNV	chr5	133478545	G	A	545	357	188	0.345	TCF7	ENSG00000081059	NON_SYNONYMOUS_CODING	ENST00000517799.1:c.223G>A	ENSP00000427968.1:p.Ala75Thr	2/6	-	-	-
S38	SNV	chr5	133478545	G	A	545	357	188	0.345	TCF7	ENSG00000081059	NON_SYNONYMOUS_CODING	ENST00000520958.1:c.544G>A	ENSP00000429547.1:p.Ala182Thr	5/8	-	-	-
S38	SNV	chr5	133478545	G	A	545	357	188	0.345	TCF7	ENSG00000081059	NON_SYNONYMOUS_CODING	ENST00000517855.1:c.254G>A	ENSP00000429178.1:p.Ala86Thr	2/6	-	-	-
S38	SNV	chr5	133478545	G	A	545	357	188	0.345	TCF7	ENSG00000081059	NON_SYNONYMOUS_CODING	ENST00000321584.4:c.889G>A	ENSP00000326540.4:p.Ala297Thr	7/10	-	-	-
S51	SNV	chr5	133482004	G	T	469	228	241	0.514	TCF7	ENSG00000081059	NON_SYNONYMOUS_CODING	ENST00000432532.2:c.893G>T	ENSP00000397946.2:p.Gly298Val	10/10	-	-	-
S51	SNV	chr5	133482004	G	T	469	228	241	0.514	TCF7	ENSG00000081059	NON_SYNONYMOUS_CODING	ENST00000395029.1:c.1238G>T	ENSP00000378472.1:p.Gly413Val	11/11	-	-	-
28	SNV	chr10	114925491	C	G	171	82	89	0.517	TCF7L2	ENSG00000148737	UTR_3_PRIME	ENST00000355717.4:c.*98C>G	-	13/13	-	0.0002	rs76249005
28	SNV	chr10	114925491	C	G	171	82	89	0.517	TCF7L2	ENSG00000148737	UTR_3_PRIME	ENST00000538897.1:c.*98C>G	-	14/14	-	0.0002	rs76249005
28	SNV	chr10	114925491	C	G	171	82	89	0.517	TCF7L2	ENSG00000148737	UTR_3_PRIME	ENST00000369386.1:c.*208C>G	-	7/7	-	0.0002	rs76249005
S17	SNV	chr14	96180397	C	T	448	230	218	0.487	TCL1A	ENSG00000100721	NON_SYNONYMOUS_CODING	ENST00000555450.1:c.7G>A	ENSP00000450701.1:p.Glu3Lys	1/4	-	-	-
S17	SNV	chr14	96180397	C	T	448	230	218	0.487	TCL1A	ENSG00000100721	NON_SYNONYMOUS_CODING	ENST00000554012.1:c.7G>A	ENSP00000451506.1:p.Glu3Lys	1/4	-	-	-
S17	SNV	chr14	96180397	C	T	448	230	218	0.487	TCL1A	ENSG00000100721	NON_SYNONYMOUS_CODING	ENST00000402399.1:c.7G>A	ENSP00000385036.1:p.Glu3Lys	1/4	YES	-	-
S17	SNV	chr14	96180397	C	T	448	230	218	0.487	TCL1A	ENSG00000100721	NON_SYNONYMOUS_CODING	ENST00000555202.1:c.7G>A	ENSP00000450496.1:p.Glu3Lys	1/3	-	-	-
S35	SNV	chr10	70446394	T	G	588	308	280	0.475	TET1	ENSG00000138336	NON_SYNONYMOUS_CODING	ENST00000373644.4:c.5334T>G	ENSP00000362748.4:p.Ile1778Met	11/12	YES	-	rs539035050
11	INDEL	chr4	106155606	T	+A	167	99	68	0.407	TET2	ENSG00000168769	FRAME_SHIFT	-	-	3/11	-	-	-
11	INDEL	chr4	106155606	T	+A	167	99	68	0.407	TET2	ENSG00000168769	FRAME_SHIFT	-	-	3/10	-	-	-
11	INDEL	chr4	106155834	C	-A	153	101	52	0.34	TET2	ENSG00000168769	FRAME_SHIFT	ENST00000394764.1:c.737delC	ENSP00000378245.1:p.Thr246AsnfsTer4	3/3	-	-	-
11	INDEL	chr4	106155834	C	-A	153	101	52	0.34	TET2	ENSG00000168769	FRAME_SHIFT	ENST00000540549.1:c.737delC	ENSP00000442788.1:p.Thr246AsnfsTer4	3/11	YES	-	-
11	INDEL	chr4	106155834	C	-A	153	101	52	0.34	TET2	ENSG00000168769	FRAME_SHIFT	ENST00000545826.1:c.737delC	ENSP00000442867.1:p.Thr246AsnfsTer4	3/10	-	-	-
11	INDEL	chr4	106155606	T	+A	167	99	68	0.407	TET2	ENSG00000168769	FRAME_SHIFT	-	-	3/11	-	-	-
11	INDEL	chr4	106155834	C	-A	153	101	52	0.34	TET2	ENSG00000168769	FRAME_SHIFT	ENST00000513237.1:c.800delC	ENSP00000425443.1:p.Thr267AsnfsTer4	3/11	-	-	-
11	INDEL	chr4	106155606	T	+A	167	99	68	0.407	TET2	ENSG00000168769	FRAME_SHIFT	-	-	3/3	-	-	-
11	INDEL	chr4	106155834	C	-A	153	101	52	0.34	TET2	ENSG00000168769	FRAME_SHIFT	ENST00000413648.2:c.737delC	ENSP00000391448.2:p.Thr246AsnfsTer4	2/3	-	-	-
11	INDEL	chr4	106155606	T	+A	167	99	68	0.407	TET2	ENSG00000168769	FRAME_SHIFT	-	-	3/11	YES	-	-

11	INDEL	chr4	106155606	T	+A	167	99	68	0.407	TET2	ENSG00000168769	FRAME_SHIFT			3/3	-	-	-	
11	INDEL	chr4	106155834	C	-A	153	101	52	0.34	TET2	ENSG00000168769	FRAME_SHIFT	ENST00000305737.2:c.737delC	ENSP00000306705.2:p.Thr246AsnfsTer4	3/3	-	-	-	
11	INDEL	chr4	106155606	T	+A	167	99	68	0.407	TET2	ENSG00000168769	FRAME_SHIFT			2/3	-	-	-	
11	INDEL	chr4	106155834	C	-A	153	101	52	0.34	TET2	ENSG00000168769	FRAME_SHIFT	ENST00000380013.4:c.737delC	ENSP00000369351.4:p.Thr246AsnfsTer4	3/11	-	-	-	
21	SNV	chr4	106162547	G	A	177	39	138	0.78	TET2	ENSG00000168769	NON_SYNONYMOUS_CODING	ENST00000513237.1:c.3524G>A	ENSP00000425443.1:p.Gly1175Asp	4/11	-	-	COSM211646	
21	SNV	chr4	106162547	G	A	177	39	138	0.78	TET2	ENSG00000168769	NON_SYNONYMOUS_CODING	ENST00000380013.4:c.3461G>A	ENSP00000369351.4:p.Gly1154Asp	4/11	-	-	COSM211646	
21	SNV	chr4	106162547	G	A	177	39	138	0.78	TET2	ENSG00000168769	NON_SYNONYMOUS_CODING	ENST00000413648.2:c.3461G>A	ENSP00000391448.2:p.Gly1154Asp	3/3	-	-	COSM211646	
21	SNV	chr4	106162547	G	A	177	39	138	0.78	TET2	ENSG00000168769	NON_SYNONYMOUS_CODING	ENST00000540549.1:c.3461G>A	ENSP00000442788.1:p.Gly1154Asp	4/11	YES	-	COSM211646	
21	SNV	chr4	106162547	G	A	177	39	138	0.78	TET2	ENSG00000168769	UTR_3_PRIME	ENST00000394764.1:c.*3950G>A	-	3/3	-	-	COSM211646	
21	SNV	chr4	106162547	G	A	177	39	138	0.78	TET2	ENSG00000168769	UTR_3_PRIME	ENST00000305737.2:c.*3950G>A	-	3/3	-	-	COSM211646	
26	INDEL	chr4	106156935	T	-G	469	291	178	0.38	TET2	ENSG00000168769	FRAME_SHIFT	ENST00000413648.2:c.1837delG	ENSP00000391448.2:p.Leu615SerfsTer24	2/3	-	-	-	
26	INDEL	chr4	106156935	T	-G	469	291	178	0.38	TET2	ENSG00000168769	FRAME_SHIFT	ENST00000540549.1:c.1837delG	ENSP00000442788.1:p.Leu615SerfsTer24	3/11	YES	-	-	
26	INDEL	chr4	106156935	T	-G	469	291	178	0.38	TET2	ENSG00000168769	FRAME_SHIFT	ENST00000513237.1:c.1900delG	ENSP00000425443.1:p.Leu636SerfsTer24	3/11	-	-	-	
26	INDEL	chr4	106156935	T	-G	469	291	178	0.38	TET2	ENSG00000168769	FRAME_SHIFT	ENST00000545826.1:c.1837delG	ENSP00000442867.1:p.Leu615SerfsTer24	3/10	-	-	-	
26	INDEL	chr4	106156935	T	-G	469	291	178	0.38	TET2	ENSG00000168769	FRAME_SHIFT	ENST00000394764.1:c.1837delG	ENSP00000378245.1:p.Leu615SerfsTer24	3/3	-	-	-	
26	INDEL	chr4	106156935	T	-G	469	291	178	0.38	TET2	ENSG00000168769	FRAME_SHIFT	ENST00000305737.2:c.1837delG	ENSP00000306705.2:p.Leu615SerfsTer24	3/3	-	-	-	
26	INDEL	chr4	106156935	T	-G	469	291	178	0.38	TET2	ENSG00000168769	FRAME_SHIFT	ENST00000380013.4:c.1837delG	ENSP00000369351.4:p.Leu615SerfsTer24	3/11	-	-	-	
38	SNV	chr4	106190827	T	C	490	325	165	0.337	TET2	ENSG00000168769	UTR_3_PRIME	ENST00000545826.1:c.*429T>C	-	8/10	-	-	-	
38	SNV	chr4	106156304	C	G	777	416	361	0.465	TET2	ENSG00000168769	STOP_GAINED	ENST00000540549.1:c.1205C>G	ENSP00000442788.1:p.Ser402Ter	3/11	YES	-	-	
38	SNV	chr4	106156304	C	G	777	416	361	0.465	TET2	ENSG00000168769	STOP_GAINED	ENST00000394764.1:c.1205C>G	ENSP00000378245.1:p.Ser402Ter	3/3	-	-	-	
38	SNV	chr4	106156304	C	G	777	416	361	0.465	TET2	ENSG00000168769	STOP_GAINED	ENST00000305737.2:c.1205C>G	ENSP00000306705.2:p.Ser402Ter	3/3	-	-	-	
38	SNV	chr4	106156304	C	G	777	416	361	0.465	TET2	ENSG00000168769	STOP_GAINED	ENST00000545826.1:c.1205C>G	ENSP00000442867.1:p.Ser402Ter	3/10	-	-	-	
38	SNV	chr4	106156304	C	G	777	416	361	0.465	TET2	ENSG00000168769	STOP_GAINED	ENST00000413648.2:c.1205C>G	ENSP00000391448.2:p.Ser402Ter	2/3	-	-	-	
38	SNV	chr4	106190827	T	C	490	325	165	0.337	TET2	ENSG00000168769	NON_SYNONYMOUS_CODING	ENST00000380013.4:c.4105T>C	ENSP00000369351.4:p.Ser1369Pro	9/11	-	-	-	
38	SNV	chr4	106190827	T	C	490	325	165	0.337	TET2	ENSG00000168769	NON_SYNONYMOUS_CODING	ENST00000540549.1:c.4105T>C	ENSP00000442788.1:p.Ser1369Pro	9/11	YES	-	-	
38	SNV	chr4	106190827	T	C	490	325	165	0.337	TET2	ENSG00000168769	NON_SYNONYMOUS_CODING	ENST00000513237.1:c.4168T>C	ENSP00000425443.1:p.Ser1390Pro	9/11	-	-	-	
38	SNV	chr4	106156304	C	G	777	416	361	0.465	TET2	ENSG00000168769	STOP_GAINED	ENST00000513237.1:c.1268C>G	ENSP00000425443.1:p.Ser423Ter	3/11	-	-	-	
38	SNV	chr4	106156304	C	G	777	416	361	0.465	TET2	ENSG00000168769	STOP_GAINED	ENST00000380013.4:c.1205C>G	ENSP00000369351.4:p.Ser402Ter	3/11	-	-	-	
42	SNV	chr4	106193782	T	C	226	157	69	0.305	TET2	ENSG00000168769	NON_SYNONYMOUS_CODING	ENST00000380013.4:c.4244T>C	ENSP00000369351.4:p.Leu1415Pro	10/11	-	-	-	
42	SNV	chr4	106193782	T	C	226	157	69	0.305	TET2	ENSG00000168769	NON_SYNONYMOUS_CODING	ENST00000513237.1:c.4307T>C	ENSP00000425443.1:p.Leu1436Pro	10/11	-	-	-	
42	SNV	chr4	106193782	T	C	226	157	69	0.305	TET2	ENSG00000168769	NON_SYNONYMOUS_CODING	ENST00000540549.1:c.4244T>C	ENSP00000442788.1:p.Leu1415Pro	10/11	YES	-	-	
42	SNV	chr4	106193782	T	C	226	157	69	0.305	TET2	ENSG00000168769	UTR_3_PRIME	ENST00000545826.1:c.*568T>C	-	9/10	-	-	-	
47	SNV	chr4	106180838	G	A	167	88	79	0.473	TET2	ENSG00000168769	NON_SYNONYMOUS_CODING	ENST00000513237.1:c.3929G>A	ENSP00000425443.1:p.Cys1310Tyr	7/11	-	-	COSM42039,COSM87136,COSM3719258	
47	SNV	chr4	106180838	G	A	167	88	79	0.473	TET2	ENSG00000168769	UTR_3_PRIME	ENST00000545826.1:c.*190G>A	-	6/10	-	-	COSM42039,COSM87136,COSM3719258	
47	SNV	chr4	106180838	G	A	167	88	79	0.473	TET2	ENSG00000168769	NON_SYNONYMOUS_CODING	ENST00000540549.1:c.3866G>A	ENSP00000442788.1:p.Cys1289Tyr	7/11	YES	-	COSM42039,COSM87136,COSM3719258	
47	SNV	chr4	106180838	G	A	167	88	79	0.473	TET2	ENSG00000168769	NON_SYNONYMOUS_CODING	ENST00000380013.4:c.3866G>A	ENSP00000369351.4:p.Cys1289Tyr	7/11	-	-	COSM42039,COSM87136,COSM3719258	
48	SNV	chr4	106164772	C	T	125	82	43	0.344	TET2	ENSG00000168769	NON_SYNONYMOUS_CODING	ENST00000380013.4:c.3640C>T	ENSP00000369351.4:p.Arg1214Trp	6/11	-	-	-	
48	SNV	chr4	106164772	C	T	125	82	43	0.344	TET2	ENSG00000168769	NON_SYNONYMOUS_CODING	ENST00000513237.1:c.3703C>T	ENSP00000425443.1:p.Arg1235Trp	6/11	-	-	-	
48	SNV	chr4	106164772	C	T	125	82	43	0.344	TET2	ENSG00000168769	NON_SYNONYMOUS_CODING	ENST00000540549.1:c.3640C>T	ENSP00000442788.1:p.Arg1214Trp	6/11	YES	-	-	
62	SNV	chr4	106193801	C	G	209	109	100	0.476	TET2	ENSG00000168769	STOP_GAINED	ENST00000380013.4:c.4263C>G	ENSP00000369351.4:p.Tyr1421Ter	10/11	-	-	-	
62	SNV	chr4	106156729	C	T	151	71	80	0.53	TET2	ENSG00000168769	STOP_GAINED	ENST00000545826.1:c.1630C>T	ENSP00000442867.1:p.Arg544Ter	3/10	-	-	COSM41850,COSM1594211	
62	SNV	chr4	106156729	C	T	151	71	80	0.53	TET2	ENSG00000168769	STOP_GAINED	ENST00000540549.1:c.1630C>T	ENSP00000442788.1:p.Arg544Ter	3/11	YES	-	COSM41850,COSM1594211	
62	SNV	chr4	106193801	C	G	209	109	100	0.476	TET2	ENSG00000168769	UTR_3_PRIME	ENST00000545826.1:c.*587C>G	-	9/10	-	-	-	
62	SNV	chr4	106156729	C	T	151	71	80	0.53	TET2	ENSG00000168769	STOP_GAINED	ENST00000380013.4:c.1630C>T	ENSP00000369351.4:p.Arg544Ter	3/11	-	-	COSM41850,COSM1594211	
62	SNV	chr4	106156729	C	T	151	71	80	0.53	TET2	ENSG00000168769	STOP_GAINED	ENST00000413648.2:c.1630C>T	ENSP00000391448.2:p.Arg544Ter	2/3	-	-	COSM41850,COSM1594211	
62	SNV	chr4	106156729	C	T	151	71	80	0.53	TET2	ENSG00000168769	STOP_GAINED	ENST00000513237.1:c.1693C>T	ENSP00000425443.1:p.Arg565Ter	3/11	-	-	COSM41850,COSM1594211	
62	SNV	chr4	106193801	C	G	209	109	100	0.476	TET2	ENSG00000168769	STOP_GAINED	ENST00000540549.1:c.4263C>G	ENSP00000442788.1:p.Tyr1421Ter	10/11	YES	-	-	
62	SNV	chr4	106156729	C	T	151	71	80	0.53	TET2	ENSG00000168769	STOP_GAINED	ENST00000305737.2:c.1630C>T	ENSP00000306705.2:p.Arg544Ter	3/3	-	-	COSM41850,COSM1594211	
62	SNV	chr4	106156729	C	T	151	71	80	0.53	TET2	ENSG00000168769	STOP_GAINED	ENST00000394764.1:c.1630C>T	ENSP00000378245.1:p.Arg544Ter	3/3	-	-	COSM41850,COSM1594211	
62	SNV	chr4	106193801	C	G	209	109	100	0.476	TET2	ENSG00000168769	STOP_GAINED	ENST00000513237.1:c.4326C>G	ENSP00000425443.1:p.Tyr1442Ter	10/11	-	-	-	
7	SNV	chr4	106196621	C	T	937	485	452	0.482	TET2	ENSG00000168769	STOP_GAINED	ENST00000540549.1:c.4954C>T	ENSP00000442788.1:p.Gln1652Ter	11/11	YES	-	-	
7	SNV	chr4	106164767	T	C	224	146	78	0.348	TET2	ENSG00000168769	NON_SYNONYMOUS_CODING	ENST00000513237.1:c.3698T>C	ENSP00000425443.1:p.Leu1233Ser	6/11	-	-	COSM1737883	
7	SNV	chr4	106164767	T	C	224	146	78	0.348	TET2	ENSG00000168769	NON_SYNONYMOUS_CODING	ENST00000380013.4:c.3635T>C	ENSP00000369351.4:p.Leu1212Ser	6/11	-	-	COSM1737883	
7	SNV	chr4	106196621	C	T	937	485	452	0.482	TET2	ENSG00000168769	STOP_GAINED	ENST00000513237.1:c.5017C>T	ENSP00000425443.1:p.Gln1673Ter	11/11	-	-	-	
7	SNV	chr4	106196621	C	T	937	485	452	0.482	TET2	ENSG00000168769	STOP_GAINED	ENST00000380013.4:c.4954C>T	ENSP00000369351.4:p.Gln1652Ter	11/11	-	-	-	
7	SNV	chr4	106196621	C	T	937	485	452	0.482	TET2	ENSG00000168769	UTR_3_PRIME	ENST00000545826.1:c.*1278C>T	-	10/10	-	-	-	
7	SNV	chr4	106164767	T	C	224	146	78	0.348	TET2	ENSG00000168769	NON_SYNONYMOUS_CODING	ENST00000540549.1:c.3635T>C	ENSP00000442788.1:p.Leu1212Ser	6/11	YES	-	COSM1737883	
7	SNV	chr4	106164767	T	C	224	146	78	0.348	TET2	ENSG00000168769	NON_SYNONYMOUS_CODING	ENST00000545826.1:c.3544T>C	ENSP00000442867.1:p.Trp1182Arg	5/10	-	-	COSM1737883	
8	INDEL	chr4	106162543	G	+C	1032	460	572	0.554	TET2	ENSG00000168769	FRAME_SHIFT	-	-	4/11	-	-	-	
8	INDEL	chr4	106162543	G	+C	1032	460	572	0.554	TET2	ENSG00000168769	UTR_3_PRIME	-	-	3/3	-	-	-	
8	INDEL	chr4	106162543	G	+C	1032	460	572	0.554	TET2	ENSG00000168769	UTR_3_PRIME	-	-	3/3	-	-	-	
8	INDEL	chr4	106162543	G	+C	1032	460	572	0.554	TET2	ENSG00000168769	FRAME_SHIFT	-	-	6/11	YES	-	-	
8	INDEL	chr4	106162543	G	+C	1032	460	572	0.554	TET2	ENSG00000168769	FRAME_SHIFT	-	-	4/11	-	-	-	
8	INDEL	chr4	106162543	G	+C	1032	460	572	0.554	TET2	ENSG00000168769	FRAME_SHIFT	-	-	3/3	-	-	-	
S02	SNV	chr4	106197365	G	T	257	130	127	0.494	TET2	ENSG00000168769	NON_SYNONYMOUS_CODING	ENST00000380013.4:c.5698G>T	ENSP00000369351.4:p.Val1900Phe	11/11	-	-	-	
S02	SNV	chr4	106197365	G	T	257	130	127	0.494	TET2	ENSG00000168769	NON_SYNONYMOUS_CODING	ENST00000513237.1:c.5761G>T	ENSP00000425443.1:p.Val1921Phe	11/11	-	-	-	
S02	SNV	chr4	106197365	G	T	257	130	127	0.494	TET2	ENSG00000168769	NON_SYNONYMOUS_CODING	ENST00000540549.1:c.5698G>T	ENSP00000442788.1:p.Val1900Phe	11/11	YES	-	-	
S10	SNV	chr4	106157974	A	T	920	503	417	0.453	TET2	ENSG00000168769	STOP_GAINED	ENST00000394764.1:c.2875A>T	ENSP00000378245.1:p.Lys959Ter	3/3	-	-	-	
S10	SNV	chr4	106157974	A	T	920	503	417	0.453	TET2	ENSG00000168769	STOP_GAINED	ENST00000513237.1:c.2938A>T	ENSP00000425443.1:p.Lys980Ter	3/11	-	-	-	
S10	SNV	chr4	106157974	A	T	920	503	417	0.453	TET2	ENSG00000168769	STOP_GAINED	ENST00000540549.1:c.2875A>T	ENSP00000442788.1:p.Lys959Ter	3/11	YES	-	-	
S10	SNV	chr4	106157974	A	T	920	503	417	0.453	TET2	ENSG00000168769	STOP_GAINED	ENST00000380013.4:c.2875A>T	ENSP00000369351.4:p.Lys959Ter	3/11	-	-	-	
S10	SNV	chr4	106157974	A	T	920	503	417	0.453	TET2	ENSG00000168769	STOP_GAINED	ENST00000545826.1:c.2875A>T	ENSP00000442867.1:p.Lys959Ter	3/10	-	-	-	
S10	SNV	chr4	106157974	A	T	920	503	417	0.453	TET2	ENSG00000168769	STOP_GAINED	ENST00000305737.2:c.2875A>T	ENSP00000306705.2:p.Lys959Ter	3/3	-	-	-	
S10	SNV	chr4	106157974	A	T	920	503	417	0.453	TET2	ENSG00000168769	STOP_GAINED	ENST00000413648.2:c.2875A>T	ENSP00000391448.2:p.Lys959Ter	2/3	-	-	-	
S17	SNV	chr4	106156747	C	T	1223	639	584	0.478	TET2	ENSG00000168769	STOP_GAINED	ENST00000413648.2:c.1648C>T	ENSP00000391448.2:p.Arg550Ter	2/3	-	0.0004	rs572712965,COSM41644,COSM1318629	
S17	INDEL	chr4	106156923	T	-TCCAACATGCC	1115	749	366	0.328	TET2	ENSG00000168769	FRAME_SHIFT	ENST00000513237.1:c.1886delT	ENSP00000425443.1:p.Ser630ProfsTer30	3/11	-	-	COSM43465	
S17	INDEL	chr4	106156923	T	-TCCAACATGCC	1115	749	366	0.328	TET2	ENSG00000168769	FRAME_SHIFT	ENST00000380013.4:c.1825delT	ENSP00000369351.4:p.Ser609ProfsTer30	3/11	-	-	COSM43465	
S17	INDEL	chr4	106156923	T	-TCCAACATGCC	1115	749	366	0.328	TET2	ENSG00000168769	FRAME_SHIFT	ENST00000305737.2:c.1825delT	ENSP00000306705.2:p.Ser609ProfsTer30	3/3	-	-	COSM43465	
S17	INDEL	chr4	106156923	T	-TCCAACATGCC	1115	749	366	0.328	TET2	ENSG00000168769	FRAME_SHIFT	ENST00000545826.1:c.1825delT	ENSP00000442867.1:p.Ser609ProfsTer30	3/10	-	-	COSM43465	
S17	SNV	chr4	106156747	C	T	1223	639	584	0.478	TET2	ENSG00000168769	STOP_GAINED	ENST00000540549.1:c.1648C>T	ENSP00000442788.1:p.Arg550Ter	3/11	YES	0.0004	rs572712965,COSM41644,COSM1318629	
S17	SNV	chr4	106156747	C	T	1223	639	584	0.478	TET2	ENSG00000168769	STOP_GAINED	ENST00000305737.2:c.1648C>T	ENSP00000306705.2:p.Arg550Ter	3/3	-	0.0004	rs572712965,COSM41644,COSM1318629	
S17	SNV	chr4	106156747	C	T	1223	639	584	0.478	TET2	ENSG00000168769	STOP_GAINED	ENST00000545826.1:c.1648C>T	ENSP00000442867.1:p.Arg550Ter	3/10	-	0.0004	rs572712965,COSM41644,COSM1318629	
S17	INDEL	chr4	106156923	T	-TCCAACATGCC	1115	749	366	0.328	TET2	ENSG00000168769	FRAME_SHIFT	ENST00000413648.2:c.1825delT	ENSP00000391448.2:p.Ser609ProfsTer30	2/3	-	-	COSM43465	
S17	INDEL	chr4	106156923	T	-TCCAACATGCC	1115	749	366	0.328	TET2	ENSG00000168769	FRAME_SHIFT	ENST00000394764.1:c.1825delT	ENSP00000378245.1:p.Ser609ProfsTer30	3/3	-	-	COSM43465	
S17	SNV	chr4	106156747	C	T	1223	639	584	0.478	TET2	ENSG00000168769	STOP_GAINED	ENST00000513237.1:c.1711C>T	ENSP00000425443.1:p.Arg571Ter	3/11	-	0.0004	rs572712965,COSM41644,COSM1318629	
S17	SNV	chr4	106156747	C	T	1223	639	584	0.478	TET2	ENSG00000168769	STOP_GAINED	ENST00000380013.4:c.1648C>T	ENSP00000369351.4:p.Arg550Ter	3/11	-	0.0004	rs572712965,COSM41644,COSM1318629	
S17	INDEL	chr4	106156923	T	-TCCAACATGCC	1115	749	366	0.328	TET2	ENSG00000168769	FRAME_SHIFT	ENST00000540549.1:c.1825delT	ENSP00000442788.1:p.Ser609ProfsTer30	3/11	YES	-	COSM43465	
S17	SNV	chr4	106156747	C	T	1223	639	584	0.478	TET2	ENSG00000168769	STOP_GAINED	ENST00000394764.1:c.1648C>T	ENSP00000378245.1:p.Arg550Ter	3/3	-	0.0004	rs572712965,COSM41644,COSM1318629	
S25	INDEL	chr4	106158364	G	-AA	51	30	21	0.412	TET2	ENSG00000168769	FRAME_SHIFT	ENST00000380013.4:c.3266delA	ENSP00000369351.4:p.Lys1090ArgfsTer16	3/11	-	-	-	
S25	INDEL	chr4	106158364	G	-AA	51	30	21	0.412	TET2	ENSG00000168769	FRAME_SHIFT	ENST00000545826.1:c.3266delA	ENSP00000442867.1:p.Lys1090ArgfsTer16	3/10	-	-	-	
S25	INDEL	chr4	106158364	G	-AA	51	30	21	0.412	TET2	ENSG00000168769	FRAME_SHIFT	ENST00000540549.1:c.3266delA	ENSP00000442788.1:p.Lys1090ArgfsTer16	3/11	YES	-	-	
S25	INDEL	chr4	106158364	G	-AA	51	30	21	0.412	TET2	ENSG00000168769	FRAME_SHIFT	ENST00000413648.2:c.3266delA	ENSP00000391448.2:p.Lys1090ArgfsTer16	2/3	-	-	-	
S25	INDEL	chr4	106158364	G	-AA	51	30	21	0.412	TET2	ENSG00000168769	FRAME_SHIFT	ENST00000513237.1:c.3329delA	ENSP00000425443.1:p.Lys1111ArgfsTer16	3/11	-	-	-	
S25	INDEL	chr4	106158364	G	-AA	51	30	21	0.412	TET2	ENSG00000168769	FRAME_SHIFT	ENST00000305737.2:c.3266delA	ENSP00000306705.2:p.Lys1090ArgfsTer16	3/3	-	-	-	
S25	INDEL	chr4	106158364	G	-AA	51	30	21	0.412	TET2	ENSG00000168769	FRAME_SHIFT	ENST00000394764.1:c.3266delA	ENSP00000378245.1:p.Lys1090ArgfsTer16	3/3	-	-	-	
S26	INDEL	chr4	106196484	G	+T	132	80	52	0.394	TET2	ENSG00000168769	FRAME_SHIFT	-	-	11/11	-	-	-	
S26	INDEL	chr4	106196484	G	+T	132	80	52	0.394	TET2	ENSG00000168769	FRAME_SHIFT	-	-	11/11	YES	-	-	
S26	INDEL	chr4	106196484	G	+T	132	80	52	0.394	TET2	ENSG00000168769	FRAME_SHIFT	-	-	11/11	-	-	-	
S39	INDEL	chr4	106180783	T	+G	449	226	223	0.497	TET2	ENSG00000168769	FRAME_SHIFT	-	-	7/11	YES	-	COSM211720,COSM211721	

S39	INDEL	chr4	106180783	T	+G	449	226	223	0.497	TET2	ENSG00000168769	FRAME_SHIFT			7/11	-	-	COSM211720,COSM211721
S39	SNV	chr4	106196243	C	T	444	254	190	0.428	TET2	ENSG00000168769	STOP_GAINED	ENST00000380013.4:c.4576C>T	ENSP00000369351.4:p.Gln1526Ter	11/11	-	-	COSM43408
S39	INDEL	chr4	106180783	T	+G	449	226	223	0.497	TET2	ENSG00000168769	FRAME_SHIFT			7/11	-	-	COSM211720,COSM211721
S39	SNV	chr4	106196243	C	T	444	254	190	0.428	TET2	ENSG00000168769	STOP_GAINED	ENST00000513237.1:c.4639C>T	ENSP00000425443.1:p.Gln1547Ter	11/11	-	-	COSM43408
S39	SNV	chr4	106196243	C	T	444	254	190	0.428	TET2	ENSG00000168769	STOP_GAINED	ENST00000540549.1:c.4576C>T	ENSP00000442788.1:p.Gln1526Ter	11/11	YES	-	COSM43408
S46	SNV	chr4	106156358	C	G	591	343	248	0.42	TET2	ENSG00000168769	STOP_GAINED	ENST00000513237.1:c.1322C>G	ENSP00000425443.1:p.Ser441Ter	3/11	-	-	rs148345467,COSM1685372
S46	SNV	chr4	106156358	C	G	591	343	248	0.42	TET2	ENSG00000168769	STOP_GAINED	ENST00000305737.2:c.1259C>G	ENSP00000306705.2:p.Ser420Ter	3/3	-	-	rs148345467,COSM1685372
S46	SNV	chr4	106156358	C	G	591	343	248	0.42	TET2	ENSG00000168769	STOP_GAINED	ENST00000380013.4:c.1259C>G	ENSP00000369351.4:p.Ser420Ter	3/11	-	-	rs148345467,COSM1685372
S46	SNV	chr4	106156358	C	G	591	343	248	0.42	TET2	ENSG00000168769	STOP_GAINED	ENST00000545826.1:c.1259C>G	ENSP00000442867.1:p.Ser420Ter	3/10	-	-	rs148345467,COSM1685372
S46	SNV	chr4	106156358	C	G	591	343	248	0.42	TET2	ENSG00000168769	STOP_GAINED	ENST00000413648.2:c.1259C>G	ENSP00000391448.2:p.Ser420Ter	2/3	-	-	rs148345467,COSM1685372
S46	SNV	chr4	106156358	C	G	591	343	248	0.42	TET2	ENSG00000168769	STOP_GAINED	ENST00000540549.1:c.1259C>G	ENSP00000442788.1:p.Ser420Ter	3/11	YES	-	rs148345467,COSM1685372
S46	SNV	chr4	106156358	C	G	591	343	248	0.42	TET2	ENSG00000168769	STOP_GAINED	ENST00000394764.1:c.1259C>G	ENSP00000378245.1:p.Ser420Ter	3/3	-	-	rs148345467,COSM1685372
S47	INDEL	chr4	106155579	T	+A	523	267	256	0.49	TET2	ENSG00000168769	FRAME_SHIFT	-	-	3/11	-	-	-
S47	INDEL	chr4	106155579	T	+A	523	267	256	0.49	TET2	ENSG00000168769	FRAME_SHIFT	-	-	3/11	-	-	-
S47	INDEL	chr4	106155579	T	+A	523	267	256	0.49	TET2	ENSG00000168769	FRAME_SHIFT	-	-	3/3	-	-	-
S47	INDEL	chr4	106155579	T	+A	523	267	256	0.49	TET2	ENSG00000168769	FRAME_SHIFT	-	-	3/10	-	-	-
S47	INDEL	chr4	106155579	T	+A	523	267	256	0.49	TET2	ENSG00000168769	FRAME_SHIFT	-	-	3/3	-	-	-
S47	INDEL	chr4	106155579	T	+A	523	267	256	0.49	TET2	ENSG00000168769	FRAME_SHIFT	-	-	3/11	YES	-	-
S47	INDEL	chr4	106155579	T	+A	523	267	256	0.49	TET2	ENSG00000168769	FRAME_SHIFT	-	-	2/3	-	-	-
S51	SNV	chr4	106190797	C	A	507	281	226	0.446	TET2	ENSG00000168769	NON_SYNONYMOUS_CODING	ENST00000380013.4:c.4075C>A	ENSP00000369351.4:p.Arg1359Ser	9/11	-	-	COSM41649
S51	SNV	chr4	106196613	A	G	717	369	348	0.485	TET2	ENSG00000168769	NON_SYNONYMOUS_CODING	ENST00000513237.1:c.5009A>G	ENSP00000425443.1:p.Tyr1670Cys	11/11	-	-	-
S51	SNV	chr4	106196613	A	G	717	369	348	0.485	TET2	ENSG00000168769	NON_SYNONYMOUS_CODING	ENST00000380013.4:c.4946A>G	ENSP00000369351.4:p.Tyr1649Cys	11/11	-	-	-
S51	SNV	chr4	106180865	G	A	496	396	100	0.202	TET2	ENSG00000168769	NON_SYNONYMOUS_CODING	ENST00000513237.1:c.3956G>A	ENSP00000425443.1:p.Cys1319Tyr	7/11	-	-	COSM43474,COSM87138
S51	SNV	chr4	106196613	A	G	717	369	348	0.485	TET2	ENSG00000168769	NON_SYNONYMOUS_CODING	ENST00000540549.1:c.4946A>G	ENSP00000442788.1:p.Tyr1649Cys	11/11	YES	-	-
S51	INDEL	chr4	106197367	C	-TT	619	442	177	0.286	TET2	ENSG00000168769	FRAME_SHIFT	ENST00000513237.1:c.576delT	ENSP00000425443.1:p.Tyr1923ThrfsTer6	11/11	-	-	-
S51	SNV	chr4	106190797	C	A	507	281	226	0.446	TET2	ENSG00000168769	NON_SYNONYMOUS_CODING	ENST00000513237.1:c.4138C>A	ENSP00000425443.1:p.Arg1380Ser	9/11	-	-	COSM41649
S51	INDEL	chr4	106197367	C	-TT	619	442	177	0.286	TET2	ENSG00000168769	FRAME_SHIFT	ENST00000540549.1:c.5701delT	ENSP00000442788.1:p.Tyr1902ThrfsTer6	11/11	YES	-	-
S51	SNV	chr4	106180865	G	A	496	396	100	0.202	TET2	ENSG00000168769	NON_SYNONYMOUS_CODING	ENST00000380013.4:c.3893G>A	ENSP00000369351.4:p.Cys1298Tyr	7/11	-	-	COSM43474,COSM87138
S51	INDEL	chr4	106197367	C	-TT	619	442	177	0.286	TET2	ENSG00000168769	FRAME_SHIFT	ENST00000380013.4:c.5701delT	ENSP00000369351.4:p.Tyr1902ThrfsTer6	11/11	-	-	-
S51	SNV	chr4	106180865	G	A	496	396	100	0.202	TET2	ENSG00000168769	NON_SYNONYMOUS_CODING	ENST00000540549.1:c.3893G>A	ENSP00000442788.1:p.Cys1298Tyr	7/11	YES	-	COSM43474,COSM87138
S51	SNV	chr4	106190797	C	A	507	281	226	0.446	TET2	ENSG00000168769	NON_SYNONYMOUS_CODING	ENST00000540549.1:c.4075C>A	ENSP00000442788.1:p.Arg1359Ser	9/11	YES	-	COSM41649
S54	SNV	chr4	106164067	T	A	718	365	353	0.492	TET2	ENSG00000168769	NON_SYNONYMOUS_CODING	ENST00000540549.1:c.3577T>A	ENSP00000442788.1:p.Cys1193Ser	5/11	YES	-	-
S54	INDEL	chr4	106190789	C	+A	726	399	327	0.449	TET2	ENSG00000168769	FRAME_SHIFT	-	-	9/11	-	-	-
S54	SNV	chr4	106164067	T	A	718	365	353	0.492	TET2	ENSG00000168769	NON_SYNONYMOUS_CODING	ENST00000545826.1:c.3486T>A	ENSP00000442867.1:p.Asp1162Glu	4/10	-	-	-
S54	INDEL	chr4	106190789	C	+A	726	399	327	0.449	TET2	ENSG00000168769	FRAME_SHIFT	-	-	9/11	-	-	-
S54	SNV	chr4	106164067	T	A	718	365	353	0.492	TET2	ENSG00000168769	NON_SYNONYMOUS_CODING	ENST00000513237.1:c.3640T>A	ENSP00000425443.1:p.Cys1214Ser	5/11	-	-	-
S54	SNV	chr4	106164067	T	A	718	365	353	0.492	TET2	ENSG00000168769	NON_SYNONYMOUS_CODING	ENST00000380013.4:c.3577T>A	ENSP00000369351.4:p.Cys1193Ser	5/11	-	-	-
S54	INDEL	chr4	106190789	C	+A	726	399	327	0.449	TET2	ENSG00000168769	FRAME_SHIFT	-	-	9/11	YES	-	-
S56	INDEL	chr4	106157202	G	+CACAC	836	547	289	0.344	TET2	ENSG00000168769	FRAME_SHIFT	-	-	3/3	-	-	-
S56	INDEL	chr4	106157202	G	+CACAC	836	547	289	0.344	TET2	ENSG00000168769	FRAME_SHIFT	-	-	3/11	-	-	-
S56	INDEL	chr4	106157202	G	+CACAC	836	547	289	0.344	TET2	ENSG00000168769	FRAME_SHIFT	-	-	3/11	-	-	-
S56	INDEL	chr4	106157202	G	+CACAC	836	547	289	0.344	TET2	ENSG00000168769	FRAME_SHIFT	-	-	3/3	-	-	-
S56	INDEL	chr4	106157202	G	+CACAC	836	547	289	0.344	TET2	ENSG00000168769	FRAME_SHIFT	-	-	3/11	YES	-	-
S56	INDEL	chr4	106157202	G	+CACAC	836	547	289	0.344	TET2	ENSG00000168769	FRAME_SHIFT	-	-	3/10	-	-	-
S56	INDEL	chr4	106157202	G	+CACAC	836	547	289	0.344	TET2	ENSG00000168769	FRAME_SHIFT	-	-	2/3	-	-	-
S61	SNV	chr4	106190894	G	T	549	35	514	0.936	TET2	ENSG00000168769	NON_SYNONYMOUS_CODING	ENST00000513237.1:c.4235G>T	ENSP00000425443.1:p.Gly1412Val	9/11	-	-	-
S61	SNV	chr4	106190894	G	T	549	35	514	0.936	TET2	ENSG00000168769	NON_SYNONYMOUS_CODING	ENST00000540549.1:c.4172G>T	ENSP00000442788.1:p.Gly1391Val	9/11	YES	-	-
S61	SNV	chr4	106190894	G	T	549	35	514	0.936	TET2	ENSG00000168769	NON_SYNONYMOUS_CODING	ENST00000380013.4:c.4172G>T	ENSP00000369351.4:p.Gly1391Val	9/11	-	-	-
S64	SNV	chr4	106190882	A	G	605	310	295	0.488	TET2	ENSG00000168769	NON_SYNONYMOUS_CODING	ENST00000513237.1:c.4223A>G	ENSP00000425443.1:p.Asn1408Ser	9/11	-	-	-
S64	SNV	chr4	106190882	A	G	605	310	295	0.488	TET2	ENSG00000168769	NON_SYNONYMOUS_CODING	ENST00000540549.1:c.4160A>G	ENSP00000442788.1:p.Asn1387Ser	9/11	YES	-	-
S64	SNV	chr4	106164920	G	A	414	218	196	0.473	TET2	ENSG00000168769	NON_SYNONYMOUS_CODING	ENST00000513237.1:c.3851G>A	ENSP00000425443.1:p.Cys1284Tyr	6/11	-	-	-
S64	SNV	chr4	106164920	G	A	414	218	196	0.473	TET2	ENSG00000168769	NON_SYNONYMOUS_CODING	ENST00000380013.4:c.3788G>A	ENSP00000369351.4:p.Cys1263Tyr	6/11	-	-	-
S64	SNV	chr4	106164920	G	A	414	218	196	0.473	TET2	ENSG00000168769	NON_SYNONYMOUS_CODING	ENST00000540549.1:c.3788G>A	ENSP00000442788.1:p.Cys1263Tyr	6/11	YES	-	-
S64	SNV	chr4	106190882	A	G	605	310	295	0.488	TET2	ENSG00000168769	NON_SYNONYMOUS_CODING	ENST00000380013.4:c.4160A>G	ENSP00000369351.4:p.Asn1387Ser	9/11	-	-	-
S66	SNV	chr4	106164778	C	T	497	243	254	0.511	TET2	ENSG00000168769	STOP_GAINED	ENST00000380013.4:c.3646C>T	ENSP00000369351.4:p.Arg1216Ter	6/11	-	-	COSM42029
S66	SNV	chr4	106164778	C	T	497	243	254	0.511	TET2	ENSG00000168769	STOP_GAINED	ENST00000540549.1:c.3646C>T	ENSP00000442788.1:p.Arg1216Ter	6/11	YES	-	COSM42029
S66	SNV	chr4	106164778	C	T	497	243	254	0.511	TET2	ENSG00000168769	STOP_GAINED	ENST00000513237.1:c.3709C>T	ENSP00000425443.1:p.Arg1237Ter	6/11	-	-	COSM42029
35	SNV	chr19	54617953	C	T	320	176	144	0.45	TFPT	ENSG00000105619	NON_SYNONYMOUS_CODING	ENST00000391757.1:c.151G>A	ENSP00000375637.1:p.Gly51Ser	2/6	-	-	-
35	SNV	chr19	54617953	C	T	320	176	144	0.45	TFPT	ENSG00000105619	NON_SYNONYMOUS_CODING	ENST00000391759.1:c.151G>A	ENSP00000375639.1:p.Gly51Ser	2/6	YES	-	-
35	SNV	chr19	54617953	C	T	320	176	144	0.45	TFPT	ENSG00000105619	NON_SYNONYMOUS_CODING	ENST00000391758.1:c.124G>A	ENSP00000375638.1:p.Gly42Ser	2/6	-	-	-
S25	SNV	chr19	54611545	C	T	99	49	50	0.505	TFPT	ENSG00000105619	NON_SYNONYMOUS_CODING	ENST00000391758.1:c.493G>A	ENSP00000375638.1:p.Gly135Ser	5/6	-	-	-
S25	SNV	chr19	54611545	C	T	99	49	50	0.505	TFPT	ENSG00000105619	NON_SYNONYMOUS_CODING	ENST00000391759.1:c.430G>A	ENSP00000375639.1:p.Gly144Ser	5/6	YES	-	-
36	INDEL	chr3	195803980	C	-CA	315	172	143	0.454	TFRC	ENSG00000072274	UTR_5_PRIME	ENST00000392396.3:c.-10delG	-	2/19	-	-	-
36	INDEL	chr3	195803980	C	-CA	315	172	143	0.454	TFRC	ENSG00000072274	UTR_5_PRIME	ENST00000535031.1:c.-458delG	-	2/17	-	-	-
36	INDEL	chr3	195803980	C	-CA	315	172	143	0.454	TFRC	ENSG00000072274	UTR_5_PRIME	ENST00000540528.1:c.-13delG	-	2/18	-	-	-
36	INDEL	chr3	195803980	C	-CA	315	172	143	0.454	TFRC	ENSG00000072274	UTR_5_PRIME	ENST00000360110.4:c.-10delG	-	2/19	YES	-	-
36	INDEL	chr3	195803980	C	-CA	315	172	143	0.454	TFRC	ENSG00000072274	UTR_5_PRIME	ENST00000420415.1:c.-51delG	-	2/18	-	-	-
S10	SNV	chr3	195794974	T	C	698	366	332	0.476	TFRC	ENSG00000072274	NON_SYNONYMOUS_CODING	ENST00000392396.3:c.829A>G	ENSP00000376197.3:p.Ile277Val	8/19	-	0.0002	rs533268185
S10	SNV	chr3	195794974	T	C	698	366	332	0.476	TFRC	ENSG00000072274	NON_SYNONYMOUS_CODING	ENST00000420415.1:c.586A>G	ENSP00000390133.1:p.Ile196Val	7/18	-	0.0002	rs533268185
S10	SNV	chr3	195794974	T	C	698	366	332	0.476	TFRC	ENSG00000072274	NON_SYNONYMOUS_CODING	ENST00000360110.4:c.829A>G	ENSP00000353224.4:p.Ile277Val	8/19	YES	0.0002	rs533268185
S61	SNV	chr3	195785200	G	T	501	263	238	0.473	TFRC	ENSG00000072274	NON_SYNONYMOUS_CODING	ENST00000420415.1:c.1389C>A	ENSP00000390133.1:p.Phe463Leu	15/18	-	-	-
S61	SNV	chr3	195785200	G	T	501	263	238	0.473	TFRC	ENSG00000072274	NON_SYNONYMOUS_CODING	ENST00000392396.3:c.1632C>A	ENSP00000376197.3:p.Phe544Leu	16/19	-	-	-
S61	SNV	chr3	195785200	G	T	501	263	238	0.473	TFRC	ENSG00000072274	NON_SYNONYMOUS_CODING	ENST00000535031.1:c.786C>A	ENSP00000437753.1:p.Phe262Leu	14/17	-	-	-
S61	SNV	chr3	195785200	G	T	501	263	238	0.473	TFRC	ENSG00000072274	NON_SYNONYMOUS_CODING	ENST00000360110.4:c.1632C>A	ENSP00000353224.4:p.Phe544Leu	16/19	YES	-	-
7	SNV	chr1	36769633	G	A	394	222	172	0.436	THRAP3	ENSG00000054118	UTR_3_PRIME	ENST00000354618.5:c.*15G>A	-	12/12	YES	-	-
7	SNV	chr1	36769633	G	A	394	222	172	0.436	THRAP3	ENSG00000054118	UTR_3_PRIME	ENST00000469141.2:c.*15G>A	-	13/13	-	-	-
S48	SNV	chr1	36767233	A	G	599	296	303	0.506	THRAP3	ENSG00000054118	NON_SYNONYMOUS_CODING	ENST00000354618.5:c.2582A>G	ENSP00000346634.5:p.Gln861Arg	11/12	YES	-	rs374762961
S48	SNV	chr1	36767233	A	G	599	296	303	0.506	THRAP3	ENSG00000054118	NON_SYNONYMOUS_CODING	ENST00000469141.2:c.2582A>G	ENSP00000433825.1:p.Gln861Arg	12/13	-	-	rs374762961
S37	SNV	chr6	138200419	G	A	591	383	208	0.352	TNFAIP3	ENSG00000118503	NON_SYNONYMOUS_CODING	ENST00000237289.4:c.1837G>A	ENSP00000237289.4:p.Val613Met	7/9	YES	-	-
S39	SNV	chr6	138199807	G	A	498	246	252	0.506	TNFAIP3	ENSG00000118503	NON_SYNONYMOUS_CODING	ENST00000237289.4:c.1225G>A	ENSP00000237289.4:p.Glu409Lys	7/9	YES	-	-
19	SNV	chr7	5427839	G	A	98	44	54	0.551	TNRC18	ENSG00000182095	NON_SYNONYMOUS_CODING	ENST00000399537.4:c.1616C>T	ENSP00000382452.4:p.Ala539Val	5/30	-	-	-
19	SNV	chr7	5427839	G	A	98	44	54	0.551	TNRC18	ENSG00000182095	NON_SYNONYMOUS_CODING	ENST00000430969.1:c.1616C>T	ENSP00000395538.1:p.Ala539Val	5/30	YES	-	-
42	SNV	chr7	5347270	G	A	287	141	146	0.509	TNRC18	ENSG00000182095	UTR_3_PRIME	ENST00000399537.4:c.*467C>T	-	30/30	-	-	-
42	SNV	chr7	5347270	G	A	287	141	146	0.509	TNRC18	ENSG00000182095	UTR_3_PRIME	ENST00000430969.1:c.*467C>T	-	30/30	YES	-	-
7	SNV	chr7	5417040	G	A	120	67	53	0.442	TNRC18	ENSG00000182095	NON_SYNONYMOUS_CODING	ENST00000413081.1:c.629C>T	ENSP00000413911.1:p.Ser210Leu	3/5	-	-	-
7	SNV	chr7	5417040	G	A	120	67	53	0.442	TNRC18	ENSG00000182095	NON_SYNONYMOUS_CODING	ENST00000430969.1:c.2423C>T	ENSP00000395538.1:p.Ser808Leu	7/30	YES	-	-
7	SNV	chr7	5417040	G	A	120	67	53	0.442	TNRC18	ENSG00000182095	NON_SYNONYMOUS_CODING	ENST00000399537.4:c.2423C>T	ENSP00000382452.4:p.Ser808Leu	7/30	-	-	-
S38	SNV	chr7	5385403	C	T	388	191	197	0.508	TNRC18	ENSG00000182095	NON_SYNONYMOUS_CODING	ENST00000430969.1:c.5509G>A	ENSP00000395538.1:p.Glu1837Lys	18/30	YES	-	-
S38	SNV	chr7	5385403	C	T	388	191	197	0.508	TNRC18	ENSG00000182095	NON_SYNONYMOUS_CODING	ENST00000399537.4:c.5509G>A	ENSP00000382452.4:p.Glu1837Lys	18/30	-	-	-
S66	SNV	chr7	5428445	G	A	45	12	33	0.733	TNRC18	ENSG00000182095	NON_SYNONYMOUS_CODING	ENST00000399537.4:c.1010C>T	ENSP00000382452.4:p.Pro337Leu	5/30	-	0.0012	rs557962394
S66	SNV	chr7	5428445	G	A	45	12	33	0.733	TNRC18	ENSG00000182095	NON_SYNONYMOUS_CODING	ENST00000430969.1:c.1010C>T	ENSP00000395538.1:p.Pro337Leu	5/30	YES	0.0012	rs557962394
S22	SNV	chr20	3971372	A	C	566	293	273	0.482	TOP1	ENSG00000198900	NON_SYNONYMOUS_CODING	ENST00000361337.2:c.578A>C	ENSP00000354522.2:p.Lys193Thr	8/21	YES	-	-
22	SNV	chr17	7578502	A	C	131	36	95	0.725	TP53	ENSG00000141510	NON_SYNONYMOUS_CODING	ENST00000508793.1:c.428T>G	ENSP00000424104.1:p.Val143Gly	5/5	-	-	TP53_g.12416T>C,TP53_g.12416T>A,TP53_g.12416T>G,COSM44573,COSM11306,COSM45382,COSM3522705,COSM3522699,COSM179830,COSM98967,COSM3522708,COSM3522702,COSM98968,COSM3522710,COSM3522704,COSM3522707,COSM3522701,COSM3388219,COSM3522706,COSM3522700,COSM16668,39,COSM3522709,COSM8969,COSM3522703
22	SNV	chr17	7578502	A	C	131	36	95	0.725	TP53	ENSG00000141510	NON_SYNONYMOUS_CODING	ENST00000509690.1:c.32T>G	ENSP00000425104.1:p.Val11Gly	2/6	-	-	TP53_g.12416T>C,TP53_g.12416T>A,TP53_g.12416T>G,COSM44573,COSM11306,COSM45382,COSM3522705,COSM3522699,COSM179830,COSM98967,COSM3522708,COSM3522702,COSM98968,COSM3522710,COSM3522704,COSM3522707,COSM3522701,COSM3388219,COSM3522706,COSM3522700,COSM16668,39,COSM3522709,COSM8969,COSM3522703

22	SNV	chr17	7578502	A	C	131	36	95	0.725	TP53	ENSG00000141510	NON_SYNONYMOUS_CODING	ENST0000045263.2:c.428T>G	ENSP00000398846.2:p.Val143Gly	5/12	-	-	TP53_g.12416T>C,TP53_g.12416T>A,TP53_g.12416T>G,COSM44573,COSM11306,COSM45382,COSM3522705,COSM3522699,COSM179830,COSM98967,COSM3522708,COSM3522702,COSM98968,COSM3522710,COSM3522704,COSM3522707,COSM3522701,COSM3388219,COSM3522706,COSM3522700,COSM1666839,COSM3522709,COSM98969,COSM3522703
22	SNV	chr17	7578502	A	C	131	36	95	0.725	TP53	ENSG00000141510	NON_SYNONYMOUS_CODING	ENST0000042046.2:c.428T>G	ENSP00000391127.2:p.Val143Gly	5/12	-	-	TP53_g.12416T>C,TP53_g.12416T>A,TP53_g.12416T>G,COSM44573,COSM11306,COSM45382,COSM3522705,COSM3522699,COSM179830,COSM98967,COSM3522708,COSM3522702,COSM98968,COSM3522710,COSM3522704,COSM3522707,COSM3522701,COSM3388219,COSM3522706,COSM3522700,COSM1666839,COSM3522709,COSM98969,COSM3522703
22	SNV	chr17	7578502	A	C	131	36	95	0.725	TP53	ENSG00000141510	NON_SYNONYMOUS_CODING	ENST0000051944.1:c.149T>G	ENSP00000423862.1:p.Val50Gly	4/6	-	-	TP53_g.12416T>C,TP53_g.12416T>A,TP53_g.12416T>G,COSM44573,COSM11306,COSM45382,COSM3522705,COSM3522699,COSM179830,COSM98967,COSM3522708,COSM3522702,COSM98968,COSM3522710,COSM3522704,COSM3522707,COSM3522701,COSM3388219,COSM3522706,COSM3522700,COSM1666839,COSM3522709,COSM98969,COSM3522703
22	SNV	chr17	7578502	A	C	131	36	95	0.725	TP53	ENSG00000141510	NON_SYNONYMOUS_CODING	ENST0000041346 5.2:c.428T>G	ENSP00000410739.2:p.Val143Gly	4/7	-	-	TP53_g.12416T>C,TP53_g.12416T>A,TP53_g.12416T>G,COSM44573,COSM11306,COSM45382,COSM3522705,COSM3522699,COSM179830,COSM98967,COSM3522708,COSM3522702,COSM98968,COSM3522710,COSM3522704,COSM3522707,COSM3522701,COSM3388219,COSM3522706,COSM3522700,COSM1666839,COSM3522709,COSM98969,COSM3522703
22	SNV	chr17	7578502	A	C	131	36	95	0.725	TP53	ENSG00000141510	NON_SYNONYMOUS_CODING	ENST0000035975 97.4:c.428T>G	ENSP00000352610.4:p.Val143Gly	4/9	-	-	TP53_g.12416T>C,TP53_g.12416T>A,TP53_g.12416T>G,COSM44573,COSM11306,COSM45382,COSM3522705,COSM3522699,COSM179830,COSM98967,COSM3522708,COSM3522702,COSM98968,COSM3522710,COSM3522704,COSM3522707,COSM3522701,COSM3388219,COSM3522706,COSM3522700,COSM1666839,COSM3522709,COSM98969,COSM3522703
22	SNV	chr17	7578502	A	C	131	36	95	0.725	TP53	ENSG00000141510	NON_SYNONYMOUS_CODING	ENST0000045888.2:c.428T>G	ENSP00000391478.2:p.Val143Gly	5/11	-	-	TP53_g.12416T>C,TP53_g.12416T>A,TP53_g.12416T>G,COSM44573,COSM11306,COSM45382,COSM3522705,COSM3522699,COSM179830,COSM98967,COSM3522708,COSM3522702,COSM98968,COSM3522710,COSM3522704,COSM3522707,COSM3522701,COSM3388219,COSM3522706,COSM3522700,COSM1666839,COSM3522709,COSM98969,COSM3522703
22	SNV	chr17	7578502	A	C	131	36	95	0.725	TP53	ENSG00000141510	NON_SYNONYMOUS_CODING	ENST0000026930 5.4:c.428T>G	ENSP00000269305.4:p.Val143Gly	5/11	YES	-	TP53_g.12416T>C,TP53_g.12416T>A,TP53_g.12416T>G,COSM44573,COSM11306,COSM45382,COSM3522705,COSM3522699,COSM179830,COSM98967,COSM3522708,COSM3522702,COSM98968,COSM3522710,COSM3522704,COSM3522707,COSM3522701,COSM3388219,COSM3522706,COSM3522700,COSM1666839,COSM3522709,COSM98969,COSM3522703
28	SNV	chr17	7577538	C	T	211	108	103	0.488	TP53	ENSG00000141510	NON_SYNONYMOUS_CODING	ENST0000045888.2:c.743G>A	ENSP00000391478.2:p.Arg248Gln	7/11	-	-	rs11540652,TP53_g.13380G>A,TP53_g.13380G>T,TP53_g.13380G>C,TP53_g.13380del,COSM10662,COSM11491,COSM6549,COSM46135,COSM340105,COSM99602,COSM241995,COSM99020,COSM241994,COSM340106,COSM3356964,COSM3378346,COSM1640830,COSM1646857,COSM1646858,COSM99021,COSM241996,COSM340107,CM920675
28	SNV	chr17	7577538	C	T	211	108	103	0.488	TP53	ENSG00000141510	NON_SYNONYMOUS_CODING	ENST0000041346 5.2:c.743G>A	ENSP00000410739.2:p.Arg248Gln	6/7	-	-	rs11540652,TP53_g.13380G>A,TP53_g.13380G>T,TP53_g.13380G>C,TP53_g.13380del,COSM10662,COSM11491,COSM6549,COSM46135,COSM340105,COSM99602,COSM241995,COSM99020,COSM241994,COSM340106,COSM3356964,COSM3378346,COSM1640830,COSM1646857,COSM1646858,COSM99021,COSM241996,COSM340107,CM920675
28	SNV	chr17	7577538	C	T	211	108	103	0.488	TP53	ENSG00000141510	NON_SYNONYMOUS_CODING	ENST0000051944.1:c.464G>A	ENSP00000423862.1:p.Arg155Gln	6/6	-	-	rs11540652,TP53_g.13380G>A,TP53_g.13380G>T,TP53_g.13380G>C,TP53_g.13380del,COSM10662,COSM11491,COSM6549,COSM46135,COSM340105,COSM99602,COSM241995,COSM99020,COSM241994,COSM340106,COSM3356964,COSM3378346,COSM1640830,COSM1646857,COSM1646858,COSM99021,COSM241996,COSM340107,CM920675
28	SNV	chr17	7577538	C	T	211	108	103	0.488	TP53	ENSG00000141510	NON_SYNONYMOUS_CODING	ENST0000050969 0.1:c.347G>A	ENSP00000425104.1:p.Arg116Gln	4/6	-	-	rs11540652,TP53_g.13380G>A,TP53_g.13380G>T,TP53_g.13380G>C,TP53_g.13380del,COSM10662,COSM11491,COSM6549,COSM46135,COSM340105,COSM99602,COSM241995,COSM99020,COSM241994,COSM340106,COSM3356964,COSM3378346,COSM1640830,COSM1646857,COSM1646858,COSM99021,COSM241996,COSM340107,CM920675
28	SNV	chr17	7577538	C	T	211	108	103	0.488	TP53	ENSG00000141510	NON_SYNONYMOUS_CODING	ENST0000035975 97.4:c.743G>A	ENSP00000352610.4:p.Arg248Gln	6/9	-	-	rs11540652,TP53_g.13380G>A,TP53_g.13380G>T,TP53_g.13380G>C,TP53_g.13380del,COSM10662,COSM11491,COSM6549,COSM46135,COSM340105,COSM99602,COSM241995,COSM99020,COSM241994,COSM340106,COSM3356964,COSM3378346,COSM1640830,COSM1646857,COSM1646858,COSM99021,COSM241996,COSM340107,CM920675
28	SNV	chr17	7577538	C	T	211	108	103	0.488	TP53	ENSG00000141510	NON_SYNONYMOUS_CODING	ENST0000026930 5.4:c.743G>A	ENSP00000269305.4:p.Arg248Gln	7/11	YES	-	rs11540652,TP53_g.13380G>A,TP53_g.13380G>T,TP53_g.13380G>C,TP53_g.13380del,COSM10662,COSM11491,COSM6549,COSM46135,COSM340105,COSM99602,COSM241995,COSM99020,COSM241994,COSM340106,COSM3356964,COSM3378346,COSM1640830,COSM1646857,COSM1646858,COSM99021,COSM241996,COSM340107,CM920675
28	SNV	chr17	7577538	C	T	211	108	103	0.488	TP53	ENSG00000141510	NON_SYNONYMOUS_CODING	ENST0000042046.2:c.743G>A	ENSP00000391127.2:p.Arg248Gln	7/12	-	-	rs11540652,TP53_g.13380G>A,TP53_g.13380G>T,TP53_g.13380G>C,TP53_g.13380del,COSM10662,COSM11491,COSM6549,COSM46135,COSM340105,COSM99602,COSM241995,COSM99020,COSM241994,COSM340106,COSM3356964,COSM3378346,COSM1640830,COSM1646857,COSM1646858,COSM99021,COSM241996,COSM340107,CM920675
28	SNV	chr17	7577538	C	T	211	108	103	0.488	TP53	ENSG00000141510	NON_SYNONYMOUS_CODING	ENST0000045263.2:c.743G>A	ENSP00000398846.2:p.Arg248Gln	7/12	-	-	rs11540652,TP53_g.13380G>A,TP53_g.13380G>T,TP53_g.13380G>C,TP53_g.13380del,COSM10662,COSM11491,COSM6549,COSM46135,COSM340105,COSM99602,COSM241995,COSM99020,COSM241994,COSM340106,COSM3356964,COSM3378346,COSM1640830,COSM1646857,COSM1646858,COSM99021,COSM241996,COSM340107,CM920675
55	SNV	chr17	7577022	G	A	416	33	383	0.921	TP53	ENSG00000141510	STOP_GAINED	ENST0000042046.2:c.916C>T	ENSP00000391127.2:p.Arg306Ter	8/12	-	-	rs121913344,TP53_g.13896C>T,TP53_g.13896del,COSM10663,COSM44631,COSM99947,COSM3388168,COSM1640820,CM971506
55	SNV	chr17	7577022	G	A	416	33	383	0.921	TP53	ENSG00000141510	STOP_GAINED	ENST0000050969 0.1:c.520C>T	ENSP00000425104.1:p.Arg174Ter	5/6	-	-	rs121913344,TP53_g.13896C>T,TP53_g.13896del,COSM10663,COSM44631,COSM99947,COSM3388168,COSM1640820,CM971506
55	SNV	chr17	7577022	G	A	416	33	383	0.921	TP53	ENSG00000141510	STOP_GAINED	ENST0000044588 8.2:c.916C>T	ENSP00000391478.2:p.Arg306Ter	8/11	-	-	rs121913344,TP53_g.13896C>T,TP53_g.13896del,COSM10663,COSM44631,COSM99947,COSM3388168,COSM1640820,CM971506
55	SNV	chr17	7577022	G	A	416	33	383	0.921	TP53	ENSG00000141510	STOP_GAINED	ENST0000045526 3.2:c.916C>T	ENSP00000398846.2:p.Arg306Ter	8/12	-	-	rs121913344,TP53_g.13896C>T,TP53_g.13896del,COSM10663,COSM44631,COSM99947,COSM3388168,COSM1640820,CM971506
55	SNV	chr17	7577022	G	A	416	33	383	0.921	TP53	ENSG00000141510	STOP_GAINED	ENST0000026930 5.4:c.916C>T	ENSP00000269305.4:p.Arg306Ter	8/11	YES	-	rs121913344,TP53_g.13896C>T,TP53_g.13896del,COSM10663,COSM44631,COSM99947,COSM3388168,COSM1640820,CM971506
55	SNV	chr17	7577022	G	A	416	33	383	0.921	TP53	ENSG00000141510	STOP_GAINED	ENST0000035975 97.4:c.916C>T	ENSP00000352610.4:p.Arg306Ter	7/9	-	-	rs121913344,TP53_g.13896C>T,TP53_g.13896del,COSM10663,COSM44631,COSM99947,COSM3388168,COSM1640820,CM971506
64	SNV	chr17	7578535	T	G	93	66	27	0.29	TP53	ENSG00000141510	NON_SYNONYMOUS_CODING	ENST0000042046.2:c.395A>C	ENSP00000391127.2:p.Lys132Thr	5/12	-	-	TP53_g.12383A>G,TP53_g.12383A>T,TP53_g.12383A>C,COSM11582,COSM43592,COSM43912,COSM179832,COSM308311,COSM308310,COSM179831,COSM3388223,COSM3388224,COSM1646844,COSM2744954,COSM308312,COSM179833
64	SNV	chr17	7578535	T	G	93	66	27	0.29	TP53	ENSG00000141510	UTR_5_PRIME	ENST0000050969 0.1:c.-2A>C	-	2/6	-	-	TP53_g.12383A>G,TP53_g.12383A>T,TP53_g.12383A>C,COSM11582,COSM43592,COSM43912,COSM179832,COSM308311,COSM308310,COSM179831,COSM3388223,COSM3388224,COSM1646844,COSM2744954,COSM308312,COSM179833
64	SNV	chr17	7578535	T	G	93	66	27	0.29	TP53	ENSG00000141510	NON_SYNONYMOUS_CODING	ENST0000026930 5.4:c.395A>C	ENSP00000269305.4:p.Lys132Thr	5/11	YES	-	TP53_g.12383A>G,TP53_g.12383A>T,TP53_g.12383A>C,COSM11582,COSM43592,COSM43912,COSM179832,COSM308311,COSM308310,COSM179831,COSM3388223,COSM3388224,COSM1646844,COSM2744954,COSM308312,COSM179833
64	SNV	chr17	7578535	T	G	93	66	27	0.29	TP53	ENSG00000141510	NON_SYNONYMOUS_CODING	ENST0000045526 3.2:c.395A>C	ENSP00000398846.2:p.Lys132Thr	5/12	-	-	TP53_g.12383A>G,TP53_g.12383A>T,TP53_g.12383A>C,COSM11582,COSM43592,COSM43912,COSM179832,COSM308311,COSM308310,COSM179831,COSM3388223,COSM3388224,COSM1646844,COSM2744954,COSM308312,COSM179833
64	SNV	chr17	7578535	T	G	93	66	27	0.29	TP53	ENSG00000141510	NON_SYNONYMOUS_CODING	ENST0000050879 3.1:c.395A>C	ENSP00000424104.1:p.Lys132Thr	5/5	-	-	TP53_g.12383A>G,TP53_g.12383A>T,TP53_g.12383A>C,COSM11582,COSM43592,COSM43912,COSM179832,COSM308311,COSM308310,COSM179831,COSM3388223,COSM3388224,COSM1646844,COSM2744954,COSM308312,COSM179833
64	SNV	chr17	7578535	T	G	93	66	27	0.29	TP53	ENSG00000141510	NON_SYNONYMOUS_CODING	ENST0000044588 8.2:c.395A>C	ENSP00000391478.2:p.Lys132Thr	5/11	-	-	TP53_g.12383A>G,TP53_g.12383A>T,TP53_g.12383A>C,COSM11582,COSM43592,COSM43912,COSM179832,COSM308311,COSM308310,COSM179831,COSM3388223,COSM3388224,COSM1646844,COSM2744954,COSM308312,COSM179833
64	SNV	chr17	7578535	T	G	93	66	27	0.29	TP53	ENSG00000141510	NON_SYNONYMOUS_CODING	ENST0000035975 97.4:c.395A>C	ENSP00000352610.4:p.Lys132Thr	4/9	-	-	TP53_g.12383A>G,TP53_g.12383A>T,TP53_g.12383A>C,COSM11582,COSM43592,COSM43912,COSM179832,COSM308311,COSM308310,COSM179831,COSM3388223,COSM3388224,COSM1646844,COSM2744954,COSM308312,COSM179833
64	SNV	chr17	7578535	T	G	93	66	27	0.29	TP53	ENSG00000141510	NON_SYNONYMOUS_CODING	ENST0000041346 5.2:c.395A>C	ENSP00000410739.2:p.Lys132Thr	4/7	-	-	TP53_g.12383A>G,TP53_g.12383A>T,TP53_g.12383A>C,COSM11582,COSM43592,COSM43912,COSM179832,COSM308311,COSM308310,COSM179831,COSM3388223,COSM3388224,COSM1646844,COSM2744954,COSM308312,COSM179833
64	SNV	chr17	7578535	T	G	93	66	27	0.29	TP53	ENSG00000141510	NON_SYNONYMOUS_CODING	ENST0000051944.1:c.116A>C	ENSP00000423862.1:p.Lys39Thr	4/6	-	-	TP53_g.12383A>G,TP53_g.12383A>T,TP53_g.12383A>C,COSM11582,COSM43592,COSM43912,COSM179832,COSM308311,COSM308310,COSM179831,COSM3388223,COSM3388224,COSM1646844,COSM2744954,COSM308312,COSM179833
8	SNV	chr17	7577548	C	G	396	115	281	0.71	TP53	ENSG00000141510	NON_SYNONYMOUS_CODING	ENST0000042046.2:c.733G>C	ENSP00000391127.2:p.Gly245Arg	7/12	-	-	TP53_g.13370G>A,TP53_g.13370G>T,TP53_g.13370G>C,TP53_g.13370del,COSM6932,COSM11081,COSM10957,COSM44642,COSM121035,COSM562651,COSM1610835,COSM121036,COSM562652,COSM1611836,COSM3356965,COSM3388190,COSM3717640,COSM3717639,COSM1640833,COSM1646855,COSM121037,COSM562653,COSM1610837,CM010463,CM900210,CM920674
8	SNV	chr17	7577548	C	G	396	115	281	0.71	TP53	ENSG00000141510	NON_SYNONYMOUS_CODING	ENST0000050969 0.1:c.337G>C	ENSP00000425104.1:p.Gly113Arg	4/6	-	-	TP53_g.13370G>A,TP53_g.13370G>T,TP53_g.13370G>C,TP53_g.13370del,COSM6932,COSM11081,COSM10957,COSM44642,COSM121035,COSM562651,COSM1610835,COSM121036,COSM562652,COSM1611836,COSM3356965,COSM3388190,COSM3717640,COSM3717639,

																		COSM1640833,COSM1646855,COSM121037,COSM562653,COSM1610837,CM010463,CM900210,CM920674
8	SNV	chr17	7577548	C	G	396	115	281	0.71	TP53	ENSG00000141510	NON_SYNONYMOUS_CODING	ENST00000359597.4:c.733G>C	ENSP00000352610.4:p.Gly245Arg	6/9	-	-	TP53_g.13370G>A,TP53_g.13370G>T,TP53_g.13370G>C,TP53_g.13370del,COSM6932,COSM11081,COSM10957,COSM44642,COSM121035,COSM562651,COSM1610835,COSM121036,COSM562652,COSM1610836,COSM3356965,COSM3388190,COSM3717640,COSM3717639,COSM1640833,COSM1646855,COSM121037,COSM562653,COSM1610837,CM010463,CM900210,CM920674
8	SNV	chr17	7577548	C	G	396	115	281	0.71	TP53	ENSG00000141510	NON_SYNONYMOUS_CODING	ENST00000455263.2:c.733G>C	ENSP00000398846.2:p.Gly245Arg	7/12	-	-	TP53_g.13370G>A,TP53_g.13370G>T,TP53_g.13370G>C,TP53_g.13370del,COSM6932,COSM11081,COSM10957,COSM44642,COSM121035,COSM562651,COSM1610835,COSM121036,COSM562652,COSM1610836,COSM3356965,COSM3388190,COSM3717640,COSM3717639,COSM1640833,COSM1646855,COSM121037,COSM562653,COSM1610837,CM010463,CM900210,CM920674
8	SNV	chr17	7577548	C	G	396	115	281	0.71	TP53	ENSG00000141510	NON_SYNONYMOUS_CODING	ENST00000514944.1:c.454G>C	ENSP00000423862.1:p.Gly152Arg	6/6	-	-	TP53_g.13370G>A,TP53_g.13370G>T,TP53_g.13370G>C,TP53_g.13370del,COSM6932,COSM11081,COSM10957,COSM44642,COSM121035,COSM562651,COSM1610835,COSM121036,COSM562652,COSM1610836,COSM3356965,COSM3388190,COSM3717640,COSM3717639,COSM1640833,COSM1646855,COSM121037,COSM562653,COSM1610837,CM010463,CM900210,CM920674
8	SNV	chr17	7577548	C	G	396	115	281	0.71	TP53	ENSG00000141510	NON_SYNONYMOUS_CODING	ENST00000269305.4:c.733G>C	ENSP00000269305.4:p.Gly245Arg	7/11	YES	-	TP53_g.13370G>A,TP53_g.13370G>T,TP53_g.13370G>C,TP53_g.13370del,COSM6932,COSM11081,COSM10957,COSM44642,COSM121035,COSM562651,COSM1610835,COSM121036,COSM562652,COSM1610836,COSM3356965,COSM3388190,COSM3717640,COSM3717639,COSM1640833,COSM1646855,COSM121037,COSM562653,COSM1610837,CM010463,CM900210,CM920674
8	SNV	chr17	7577548	C	G	396	115	281	0.71	TP53	ENSG00000141510	NON_SYNONYMOUS_CODING	ENST00000413465.2:c.733G>C	ENSP00000410739.2:p.Gly245Arg	6/7	-	-	TP53_g.13370G>A,TP53_g.13370G>T,TP53_g.13370G>C,TP53_g.13370del,COSM6932,COSM11081,COSM10957,COSM44642,COSM121035,COSM562651,COSM1610835,COSM121036,COSM562652,COSM1610836,COSM3356965,COSM3388190,COSM3717640,COSM3717639,COSM1640833,COSM1646855,COSM121037,COSM562653,COSM1610837,CM010463,CM900210,CM920674
8	SNV	chr17	7577548	C	G	396	115	281	0.71	TP53	ENSG00000141510	NON_SYNONYMOUS_CODING	ENST00000445888.2:c.733G>C	ENSP00000391478.2:p.Gly245Arg	7/11	-	-	TP53_g.13370G>A,TP53_g.13370G>T,TP53_g.13370G>C,TP53_g.13370del,COSM6932,COSM11081,COSM10957,COSM44642,COSM121035,COSM562651,COSM1610835,COSM121036,COSM562652,COSM1610836,COSM3356965,COSM3388190,COSM3717640,COSM3717639,COSM1640833,COSM1646855,COSM121037,COSM562653,COSM1610837,CM010463,CM900210,CM920674
S05	SNV	chr17	7577114	C	T	114	21	93	0.816	TP53	ENSG00000141510	NON_SYNONYMOUS_CODING	ENST00000359597.4:c.824G>A	ENSP00000352610.4:p.Cys275Tyr	7/9	-	-	TP53_g.13804G>T,TP53_g.13804G>A,TP53_g.13804G>C,TP53_g.13804del,COSM10701,COSM10893,COSM45413,COSM44178,COSM165084,COSM99932,COSM1167915,COSM318164,COSM3403255,COSM3723938,COSM1637959,COSM2744531,COSM1728541,CM076568,CM951234
S05	SNV	chr17	7577114	C	T	114	21	93	0.816	TP53	ENSG00000141510	NON_SYNONYMOUS_CODING	ENST00000269305.4:c.824G>A	ENSP00000269305.4:p.Cys275Tyr	8/11	YES	-	TP53_g.13804G>T,TP53_g.13804G>A,TP53_g.13804G>C,TP53_g.13804del,COSM10701,COSM10893,COSM45413,COSM44178,COSM165084,COSM99932,COSM1167915,COSM318164,COSM3403255,COSM3723938,COSM1637959,COSM2744531,COSM1728541,CM076568,CM951234
S05	SNV	chr17	7577114	C	T	114	21	93	0.816	TP53	ENSG00000141510	NON_SYNONYMOUS_CODING	ENST00000445888.2:c.824G>A	ENSP00000391478.2:p.Cys275Tyr	8/11	-	-	TP53_g.13804G>T,TP53_g.13804G>A,TP53_g.13804G>C,TP53_g.13804del,COSM10701,COSM10893,COSM45413,COSM44178,COSM165084,COSM99932,COSM1167915,COSM318164,COSM3403255,COSM3723938,COSM1637959,COSM2744531,COSM1728541,CM076568,CM951234
S05	SNV	chr17	7577114	C	T	114	21	93	0.816	TP53	ENSG00000141510	NON_SYNONYMOUS_CODING	ENST00000420246.2:c.824G>A	ENSP00000391127.2:p.Cys275Tyr	8/12	-	-	TP53_g.13804G>T,TP53_g.13804G>A,TP53_g.13804G>C,TP53_g.13804del,COSM10701,COSM10893,COSM45413,COSM44178,COSM165084,COSM99932,COSM1167915,COSM318164,COSM3403255,COSM3723938,COSM1637959,COSM2744531,COSM1728541,CM076568,CM951234
S05	SNV	chr17	7577114	C	T	114	21	93	0.816	TP53	ENSG00000141510	NON_SYNONYMOUS_CODING	ENST00000509690.1:c.428G>A	ENSP00000425104.1:p.Cys143Tyr	5/6	-	-	TP53_g.13804G>T,TP53_g.13804G>A,TP53_g.13804G>C,TP53_g.13804del,COSM10701,COSM10893,COSM45413,COSM44178,COSM165084,COSM99932,COSM1167915,COSM318164,COSM3403255,COSM3723938,COSM1637959,COSM2744531,COSM1728541,CM076568,CM951234
S05	SNV	chr17	7577114	C	T	114	21	93	0.816	TP53	ENSG00000141510	NON_SYNONYMOUS_CODING	ENST00000455263.2:c.824G>A	ENSP00000398846.2:p.Cys275Tyr	8/12	-	-	TP53_g.13804G>T,TP53_g.13804G>A,TP53_g.13804G>C,TP53_g.13804del,COSM10701,COSM10893,COSM45413,COSM44178,COSM165084,COSM99932,COSM1167915,COSM318164,COSM3403255,COSM3723938,COSM1637959,COSM2744531,COSM1728541,CM076568,CM951234
S16	SNV	chr17	7577120	C	T	331	113	218	0.659	TP53	ENSG00000141510	NON_SYNONYMOUS_CODING	ENST00000420246.2:c.818G>A	ENSP00000391127.2:p.Arg273His	8/12	-	0.0002	rs28934576,TP53_g.13798G>A,TP53_g.13798G>T,TP53_g.13798G>C,TP53_g.13798del,COSM10660,COSM43896,COSM44440,COSM10779,COSM318169,COSM99729,COSM165077,COSM3356963,COSM3675521,COSM1640828,COSM1645335,COSM1646808,CM920677,CM010472,CM004342
S16	SNV	chr17	7577120	C	T	331	113	218	0.659	TP53	ENSG00000141510	NON_SYNONYMOUS_CODING	ENST00000455263.2:c.818G>A	ENSP00000398846.2:p.Arg273His	8/12	-	0.0002	rs28934576,TP53_g.13798G>A,TP53_g.13798G>T,TP53_g.13798G>C,TP53_g.13798del,COSM10660,COSM43896,COSM44440,COSM10779,COSM318169,COSM99729,COSM165077,COSM3356963,COSM3675521,COSM1640828,COSM1645335,COSM1646808,CM920677,CM010472,CM004342
S16	SNV	chr17	7577120	C	T	331	113	218	0.659	TP53	ENSG00000141510	NON_SYNONYMOUS_CODING	ENST00000359597.4:c.818G>A	ENSP00000352610.4:p.Arg273His	7/9	-	0.0002	rs28934576,TP53_g.13798G>A,TP53_g.13798G>T,TP53_g.13798G>C,TP53_g.13798del,COSM10660,COSM43896,COSM44440,COSM10779,COSM318169,COSM99729,COSM165077,COSM3356963,COSM3675521,COSM1640828,COSM1645335,COSM1646808,CM920677,CM010472,CM004342
S16	SNV	chr17	7577120	C	T	331	113	218	0.659	TP53	ENSG00000141510	NON_SYNONYMOUS_CODING	ENST00000509690.1:c.422G>A	ENSP00000425104.1:p.Arg141His	5/6	-	0.0002	rs28934576,TP53_g.13798G>A,TP53_g.13798G>T,TP53_g.13798G>C,TP53_g.13798del,COSM10660,COSM43896,COSM44440,COSM10779,COSM318169,COSM99729,COSM165077,COSM3356963,COSM3675521,COSM1640828,COSM1645335,COSM1646808,CM920677,CM010472,CM004342
S16	SNV	chr17	7577120	C	T	331	113	218	0.659	TP53	ENSG00000141510	NON_SYNONYMOUS_CODING	ENST00000445888.2:c.818G>A	ENSP00000391478.2:p.Arg273His	8/11	-	0.0002	rs28934576,TP53_g.13798G>A,TP53_g.13798G>T,TP53_g.13798G>C,TP53_g.13798del,COSM10660,COSM43896,COSM44440,COSM10779,COSM318169,COSM99729,COSM165077,COSM3356963,COSM3675521,COSM1640828,COSM1645335,COSM1646808,CM920677,CM010472,CM004342
S16	SNV	chr17	7577120	C	T	331	113	218	0.659	TP53	ENSG00000141510	NON_SYNONYMOUS_CODING	ENST00000269305.4:c.818G>A	ENSP00000269305.4:p.Arg273His	8/11	YES	0.0002	rs28934576,TP53_g.13798G>A,TP53_g.13798G>T,TP53_g.13798G>C,TP53_g.13798del,COSM10660,COSM43896,COSM44440,COSM10779,COSM318169,COSM99729,COSM165077,COSM3356963,COSM3675521,COSM1640828,COSM1645335,COSM1646808,CM920677,CM010472,CM004342
S55	INDEL	chr17	7578242	C	-ACGCAA	407	195	212	0.521	TP53	ENSG00000141510	CODON_DELETION	ENST00000413465.2:c.607delG	ENSP00000410739.2:p.Val203TrpfsTer44	5/7	-	-	TP53_g.12675del,TP53_g.12675T>C,TP53_g.12675T>A,COSM45356,COSM45969,COSM1386724,COSM1386725,COSM1386727,COSM1386726
S55	INDEL	chr17	7578242	C	-ACGCAA	407	195	212	0.521	TP53	ENSG00000141510	CODON_DELETION	ENST00000514944.1:c.328delG	ENSP00000423862.1:p.Val110TrpfsTer44	5/6	-	-	TP53_g.12675del,TP53_g.12675T>C,TP53_g.12675T>A,COSM45356,COSM45969,COSM1386724,COSM1386725,COSM1386727,COSM1386726
S55	INDEL	chr17	7578242	C	-ACGCAA	407	195	212	0.521	TP53	ENSG00000141510	CODON_DELETION	ENST00000445888.2:c.607delG	ENSP00000391478.2:p.Val203TrpfsTer44	6/11	-	-	TP53_g.12675del,TP53_g.12675T>C,TP53_g.12675T>A,COSM45356,COSM45969,COSM1386724,COSM1386725,COSM1386727,COSM1386726
S55	INDEL	chr17	7578242	C	-ACGCAA	407	195	212	0.521	TP53	ENSG00000141510	CODON_DELETION	ENST00000509690.1:c.211delG	ENSP00000425104.1:p.Val71TrpfsTer44	3/6	-	-	TP53_g.12675del,TP53_g.12675T>C,TP53_g.12675T>A,COSM45356,COSM45969,COSM1386724,COSM1386725,COSM1386727,COSM1386726
S55	INDEL	chr17	7578242	C	-ACGCAA	407	195	212	0.521	TP53	ENSG00000141510	CODON_DELETION	ENST00000455263.2:c.607delG	ENSP00000398846.2:p.Val203TrpfsTer44	6/12	-	-	TP53_g.12675del,TP53_g.12675T>C,TP53_g.12675T>A,COSM45356,COSM45969,COSM1386724,COSM1386725,COSM1386727,COSM1386726
S55	INDEL	chr17	7578242	C	-ACGCAA	407	195	212	0.521	TP53	ENSG00000141510	CODON_DELETION	ENST00000359597.4:c.607delG	ENSP00000352610.4:p.Val203TrpfsTer44	5/9	-	-	TP53_g.12675del,TP53_g.12675T>C,TP53_g.12675T>A,COSM45356,COSM45969,COSM1386724,COSM1386725,COSM1386727,COSM1386726
S55	INDEL	chr17	7578242	C	-ACGCAA	407	195	212	0.521	TP53	ENSG00000141510	CODON_DELETION	ENST00000269305.4:c.607delG	ENSP00000269305.4:p.Val203TrpfsTer44	6/11	YES	-	TP53_g.12675del,TP53_g.12675T>C,TP53_g.12675T>A,COSM45356,COSM45969,COSM1386724,COSM1386725,COSM1386727,COSM1386726
S55	INDEL	chr17	7578242	C	-ACGCAA	407	195	212	0.521	TP53	ENSG00000141510	CODON_DELETION	ENST00000420246.2:c.607delG	ENSP00000391127.2:p.Val203TrpfsTer44	6/12	-	-	TP53_g.12675del,TP53_g.12675T>C,TP53_g.12675T>A,COSM45356,COSM45969,COSM1386724,COSM1386725,COSM1386727,COSM1386726
S60	SNV	chr17	7577538	C	T	640	74	566	0.884	TP53	ENSG00000141510	NON_SYNONYMOUS_CODING	ENST00000413465.2:c.743G>A	ENSP00000410739.2:p.Arg248Gln	6/7	-	-	rs11540652,TP53_g.13380G>A,TP53_g.13380G>T,TP53_g.13380G>C,TP53_g.13380del,COSM10662,COSM11491,COSM6549,COSM46135,COSM340105,COSM99602,COSM241995,COSM99020,COSM241994,COSM340106,COSM3356964,COSM3378346,COSM1640830,COSM1646857,COSM1646858,COSM99021,COSM24 1996,COSM340107,CM920675
S60	SNV	chr17	7577538	C	T	640	74	566	0.884	TP53	ENSG00000141510	NON_SYNONYMOUS_CODING	ENST00000455263.2:c.743G>A	ENSP00000398846.2:p.Arg248Gln	7/12	-	-	rs11540652,TP53_g.13380G>A,TP53_g.13380G>T,TP53_g.13380G>C,TP53_g.13380del,COSM10662,COSM11491,COSM6549,COSM46135,COSM340105,COSM99602,COSM241995,COSM99020,COSM241994,COSM340106,COSM3356964,COSM3378346,COSM1640830,COSM1646857,COSM1646858,COSM99021,COSM24 1996,COSM340107,CM920675
S60	SNV	chr17	7577538	C	T	640	74	566	0.884	TP53	ENSG00000141510	NON_SYNONYMOUS_CODING	ENST00000509690.1:c.347G>A	ENSP00000425104.1:p.Arg116Gln	4/6	-	-	rs11540652,TP53_g.13380G>A,TP53_g.13380G>T,TP53_g.13380G>C,TP53_g.13380del,COSM10662,COSM11491,COSM6549,COSM46135,COSM340105,COSM99602,COSM241995,COSM99020,COSM241994,COSM340106,COSM3356964,COSM3378346,COSM1640830,COSM1646857,COSM1646858,COSM99021,COSM24 1996,COSM340107,CM920675

S60	SNV	chr17	7577538	C	T	640	74	566	0.884	TP53	ENSG00000141510	NON_SYNONYMOUS_CODING	ENST00000359597.4:c.743G>A	ENSP00000352610.4:p.Arg248Gln	6/9	-	-	rs11540652,TP53_g.13380G>A,TP53_g.13380G>T,TP53_g.13380G>C,TP53_g.13380del,COSM10662,COSM11491,COSM6549,COSM46135,COSM340105,COSM99602,COSM241995,COSM99020,COSM241994,COSM340106,COSM3356964,COSM3378346,COSM1640830,COSM1646857,COSM1646858,COSM99021,COSM241996,COSM340107,CM920675
S60	SNV	chr17	7577538	C	T	640	74	566	0.884	TP53	ENSG00000141510	NON_SYNONYMOUS_CODING	ENST00000445888.2:c.743G>A	ENSP00000391478.2:p.Arg248Gln	7/11	-	-	rs11540652,TP53_g.13380G>A,TP53_g.13380G>T,TP53_g.13380G>C,TP53_g.13380del,COSM10662,COSM11491,COSM6549,COSM46135,COSM340105,COSM99602,COSM241995,COSM99020,COSM241994,COSM340106,COSM3356964,COSM3378346,COSM1640830,COSM1646857,COSM1646858,COSM99021,COSM241996,COSM340107,CM920675
S60	SNV	chr17	7577538	C	T	640	74	566	0.884	TP53	ENSG00000141510	NON_SYNONYMOUS_CODING	ENST00000269305.4:c.743G>A	ENSP00000269305.4:p.Arg248Gln	7/11	YES	-	rs11540652,TP53_g.13380G>A,TP53_g.13380G>T,TP53_g.13380G>C,TP53_g.13380del,COSM10662,COSM11491,COSM6549,COSM46135,COSM340105,COSM99602,COSM241995,COSM99020,COSM241994,COSM340106,COSM3356964,COSM3378346,COSM1640830,COSM1646857,COSM1646858,COSM99021,COSM241996,COSM340107,CM920675
S60	SNV	chr17	7577538	C	T	640	74	566	0.884	TP53	ENSG00000141510	NON_SYNONYMOUS_CODING	ENST00000514944.1:c.464G>A	ENSP00000423862.1:p.Arg155Gln	6/6	-	-	rs11540652,TP53_g.13380G>A,TP53_g.13380G>T,TP53_g.13380G>C,TP53_g.13380del,COSM10662,COSM11491,COSM6549,COSM46135,COSM340105,COSM99602,COSM241995,COSM99020,COSM241994,COSM340106,COSM3356964,COSM3378346,COSM1640830,COSM1646857,COSM1646858,COSM99021,COSM241996,COSM340107,CM920675
S60	SNV	chr17	7577538	C	T	640	74	566	0.884	TP53	ENSG00000141510	NON_SYNONYMOUS_CODING	ENST00000420246.2:c.743G>A	ENSP00000391127.2:p.Arg248Gln	7/12	-	-	rs11540652,TP53_g.13380G>A,TP53_g.13380G>T,TP53_g.13380G>C,TP53_g.13380del,COSM10662,COSM11491,COSM6549,COSM46135,COSM340105,COSM99602,COSM241995,COSM99020,COSM241994,COSM340106,COSM3356964,COSM3378346,COSM1640830,COSM1646857,COSM1646858,COSM99021,COSM241996,COSM340107,CM920675
1	SNV	chr19	16186967	G	C	262	115	147	0.561	TPM4	ENSG00000167460	NON_SYNONYMOUS_CODING	ENST00000538887.1:c.225G>C	ENSP00000439135.1:p.Glu75Asp	2/10	YES	-	-
1	SNV	chr19	16186967	G	C	262	115	147	0.561	TPM4	ENSG00000167460	NON_SYNONYMOUS_CODING	ENST00000344824.6:c.225G>C	ENSP00000345230.5:p.Glu75Asp	2/9	-	-	-
34	SNV	chr1	186327772	C	T	168	99	69	0.411	TPR	ENSG00000047410	NON_SYNONYMOUS_CODING	ENST00000367478.4:c.1400G>A	ENSP00000356448.3:p.Arg467Gln	13/51	YES	-	rs377405525,COSM323954
S55	SNV	chr1	186344714	G	T	649	327	322	0.496	TPR	ENSG00000047410	NON_SYNONYMOUS_CODING	ENST00000451586.1:c.47C>A	ENSP00000408103.1:p.Ser16Tyr	1/3	-	0.0008	rs371268243
S56	SNV	chr1	186287640	T	C	984	465	519	0.527	TPR	ENSG00000047410	NON_SYNONYMOUS_CODING	ENST00000367478.4:c.6760A>G	ENSP00000356448.3:p.Thr2254Ala	48/51	YES	-	-
S25	SNV	chr10	17195634	A	C	49	38	11	0.224	TRDMT1	ENSG00000107614	SPLICE_SITE_REGION	ENST00000351358.4:c.809T>G	ENSP00000324328.7:p.Val270Gly	8/9	-	-	-
S25	SNV	chr10	17195634	A	C	49	38	11	0.224	TRDMT1	ENSG00000107614	NON_SYNONYMOUS_CODING	ENST00000412821.3:c.875T>G	ENSP00000409354.3:p.Val292Gly	9/10	-	-	-
S25	SNV	chr10	17195634	A	C	49	38	11	0.224	TRDMT1	ENSG00000107614	NON_SYNONYMOUS_CODING	ENST00000488990.1:c.578T>G	ENSP00000419625.1:p.Val193Gly	5/5	-	-	-
S25	SNV	chr10	17195634	A	C	49	38	11	0.224	TRDMT1	ENSG00000107614	NON_SYNONYMOUS_CODING	ENST00000377799.3:c.947T>G	ENSP00000367030.3:p.Val316Gly	10/11	YES	-	-
S25	SNV	chr10	17195634	A	C	49	38	11	0.224	TRDMT1	ENSG00000107614	NON_SYNONYMOUS_CODING	ENST00000457442.2:c.704T>G	ENSP00000412256.2:p.Val235Gly	8/9	-	-	-
S25	SNV	chr10	17195634	A	C	49	38	11	0.224	TRDMT1	ENSG00000107614	SPLICE_SITE_REGION	ENST00000358282.7:c.*582T>G	-	8/9	-	-	-
S25	SNV	chr10	17195633	A	C	48	37	11	0.229	TRDMT1	ENSG00000107614	SPLICE_SITE_REGION	ENST00000377799.3:c.948T>G	ENST00000377799.3:c.948T>G(p.%3D)	10/11	YES	-	-
S25	SNV	chr10	17195633	A	C	48	37	11	0.229	TRDMT1	ENSG00000107614	SPLICE_SITE_REGION	ENST00000488990.1:c.579T>G	ENST00000488990.1:c.579T>G(p.%3D)	5/5	-	-	-
S25	SNV	chr10	17195633	A	C	48	37	11	0.229	TRDMT1	ENSG00000107614	SPLICE_SITE_REGION	ENST00000457442.2:c.705T>G	ENST00000457442.2:c.705T>G(p.%3D)	8/9	-	-	-
S25	SNV	chr10	17195634	A	C	49	38	11	0.224	TRDMT1	ENSG00000107614	NON_SYNONYMOUS_CODING	ENST00000351358.4:c.809T>G	ENSP00000324328.7:p.Val270Gly	8/9	-	-	-
S25	SNV	chr10	17195634	A	C	49	38	11	0.224	TRDMT1	ENSG00000107614	SPLICE_SITE_REGION	ENST00000457442.2:c.704T>G	ENSP00000412256.2:p.Val235Gly	8/9	-	-	-
S25	SNV	chr10	17195634	A	C	49	38	11	0.224	TRDMT1	ENSG00000107614	SPLICE_SITE_REGION	ENST00000377799.3:c.947T>G	ENSP00000367030.3:p.Val316Gly	10/11	YES	-	-
S25	SNV	chr10	17195634	A	C	49	38	11	0.224	TRDMT1	ENSG00000107614	SPLICE_SITE_REGION	ENST00000412821.3:c.875T>G	ENSP00000409354.3:p.Val292Gly	9/10	-	-	-
S25	SNV	chr10	17195633	A	C	48	37	11	0.229	TRDMT1	ENSG00000107614	SPLICE_SITE_REGION	ENST00000358282.7:c.*583T>G	-	8/9	-	-	-
S25	SNV	chr10	17195634	A	C	49	38	11	0.224	TRDMT1	ENSG00000107614	SPLICE_SITE_REGION	ENST00000488990.1:c.578T>G	ENSP00000419625.1:p.Val193Gly	5/5	-	-	-
S25	SNV	chr10	17195634	A	C	49	38	11	0.224	TRDMT1	ENSG00000107614	SPLICE_SITE_REGION	ENST00000377766.5:c.*378T>G	-	7/8	-	-	-
S25	SNV	chr10	17195633	A	C	48	37	11	0.229	TRDMT1	ENSG00000107614	SPLICE_SITE_REGION	ENST00000377766.5:c.*379T>G	-	7/8	-	-	-
S25	SNV	chr10	17195633	A	C	48	37	11	0.229	TRDMT1	ENSG00000107614	SPLICE_SITE_REGION	ENST00000412821.3:c.876T>G	ENST00000412821.3:c.876T>G(p.%3D)	9/10	-	-	-
S25	SNV	chr10	17195633	A	C	48	37	11	0.229	TRDMT1	ENSG00000107614	SPLICE_SITE_REGION	ENST00000351358.4:c.810T>G	ENST00000351358.4:c.810T>G(p.%3D)	8/9	-	-	-
48	SNV	chr7	138200081	G	T	216	163	53	0.245	TRIM24	ENSG00000122779	NON_SYNONYMOUS_CODING	ENST00000343526.4:c.608G>T	ENSP00000340507.4:p.Arg203Ile	3/19	YES	-	-
48	SNV	chr7	138200081	G	T	216	163	53	0.245	TRIM24	ENSG00000122779	NON_SYNONYMOUS_CODING	ENST00000415680.2:c.608G>T	ENSP00000390829.2:p.Arg203Ile	3/19	-	-	-
S54	INDEL	chr14	92480759	C	-T	422	202	220	0.521	TRIP11	ENSG00000100815	FRAME_SHIFT	ENST00000267622.4:c.986delG	ENSP00000267622.4:p.Arg329LysfsTer4	7/21	YES	-	-
S54	INDEL	chr14	92480759	C	-T	422	202	220	0.521	TRIP11	ENSG00000100815	FRAME_SHIFT	ENST00000554357.1:c.219delG	ENSP00000451032.1:p.Arg74LysfsTer4	2/15	-	-	-
S57	SNV	chr14	92472072	G	A	1175	1	1174	0.999	TRIP11	ENSG00000100815	NON_SYNONYMOUS_CODING	ENST00000554357.1:c.1394C>T	ENSP00000451032.1:p.Arg466Cys	5/15	-	-	-
S57	SNV	chr14	92472072	G	A	1175	1	1174	0.999	TRIP11	ENSG00000100815	NON_SYNONYMOUS_CODING	ENST00000267622.4:c.2248C>T	ENSP00000267622.4:p.Arg750Cys	11/21	YES	-	-
24	SNV	chr9	135779052	G	A	51	31	20	0.392	TSC1	ENSG00000165699	NON_SYNONYMOUS_CODING	ENST00000545250.1:c.2041C>T	ENSP00000444017.1:p.His681Tyr	14/20	-	0.0014	rs118203657
24	SNV	chr9	135779052	G	A	51	31	20	0.392	TSC1	ENSG00000165699	NON_SYNONYMOUS_CODING	ENST00000298552.3:c.2194C>T	ENSP00000298552.3:p.His732Tyr	17/23	YES	0.0014	rs118203657
24	SNV	chr9	135779052	G	A	51	31	20	0.392	TSC1	ENSG00000165699	NON_SYNONYMOUS_CODING	ENST00000440111.2:c.2194C>T	ENSP00000394524.2:p.His732Tyr	15/21	-	0.0014	rs118203657
31	SNV	chr9	135779052	G	A	42	23	19	0.452	TSC1	ENSG00000165699	NON_SYNONYMOUS_CODING	ENST00000298552.3:c.2194C>T	ENSP00000298552.3:p.His732Tyr	17/23	YES	0.0014	rs118203657
31	SNV	chr9	135779052	G	A	42	23	19	0.452	TSC1	ENSG00000165699	NON_SYNONYMOUS_CODING	ENST00000545250.1:c.2041C>T	ENSP00000444017.1:p.His681Tyr	14/20	-	0.0014	rs118203657
31	SNV	chr9	135779052	G	A	42	23	19	0.452	TSC1	ENSG00000165699	NON_SYNONYMOUS_CODING	ENST00000440111.2:c.2194C>T	ENSP00000394524.2:p.His732Tyr	15/21	-	0.0014	rs118203657
S59	SNV	chr9	135779052	G	A	175	90	85	0.486	TSC1	ENSG00000165699	NON_SYNONYMOUS_CODING	ENST00000545250.1:c.2041C>T	ENSP00000444017.1:p.His681Tyr	14/20	-	0.0014	rs118203657
S59	SNV	chr9	135779052	G	A	175	90	85	0.486	TSC1	ENSG00000165699	NON_SYNONYMOUS_CODING	ENST00000298552.3:c.2194C>T	ENSP00000298552.3:p.His732Tyr	17/23	YES	0.0014	rs118203657
S59	SNV	chr9	135779052	G	A	175	90	85	0.486	TSC1	ENSG00000165699	NON_SYNONYMOUS_CODING	ENST00000440111.2:c.2194C>T	ENSP00000394524.2:p.His732Tyr	15/21	-	0.0014	rs118203657
58	SNV	chr16	2138622	C	G	295	151	144	0.488	TSC2	ENSG00000103197	UTR_3_PRIME	ENST00000219476.3:c.*11C>G	-	42/42	YES	-	-
58	SNV	chr16	2138622	C	G	295	151	144	0.488	TSC2	ENSG00000103197	UTR_3_PRIME	ENST00000353929.4:c.*11C>G	-	41/41	-	-	-
58	SNV	chr16	2138622	C	G	295	151	144	0.488	TSC2	ENSG00000103197	UTR_3_PRIME	ENST00000350773.4:c.*11C>G	-	41/41	-	-	-
58	SNV	chr16	2138622	C	G	295	151	144	0.488	TSC2	ENSG00000103197	UTR_3_PRIME	ENST00000439673.2:c.*11C>G	-	39/39	-	-	-
58	SNV	chr16	2138622	C	G	295	151	144	0.488	TSC2	ENSG00000103197	UTR_3_PRIME	ENST00000401874.2:c.*11C>G	-	40/40	-	-	-
58	SNV	chr16	2138622	C	G	295	151	144	0.488	TSC2	ENSG00000103197	UTR_3_PRIME	ENST00000382538.6:c.*11C>G	-	39/39	-	-	-
S06	SNV	chr16	2106721	C	T	497	260	237	0.476	TSC2	ENSG00000103197	NON_SYNONYMOUS_CODING	ENST00000219476.3:c.725C>T	ENSP00000219476.3:p.Thr242Ile	8/42	YES	-	-
S06	SNV	chr16	2106721	C	T	497	260	237	0.476	TSC2	ENSG00000103197	NON_SYNONYMOUS_CODING	ENST00000467949.1:c.161C>T	ENSP00000454997.1:p.Thr54Ile	2/6	-	-	-
S06	SNV	chr16	2106721	C	T	497	260	237	0.476	TSC2	ENSG00000103197	NON_SYNONYMOUS_CODING	ENST00000568454.1:c.758C>T	ENSP00000454487.1:p.Thr253Ile	8/40	-	-	-
S06	SNV	chr16	2106721	C	T	497	260	237	0.476	TSC2	ENSG00000103197	NON_SYNONYMOUS_CODING	ENST00000353929.4:c.725C>T	ENSP00000248099.4:p.Thr242Ile	8/41	-	-	-
S06	SNV	chr16	2106721	C	T	497	260	237	0.476	TSC2	ENSG00000103197	NON_SYNONYMOUS_CODING	ENST00000382538.6:c.578C>T	ENSP00000371978.6:p.Thr193Ile	7/39	-	-	-
S06	SNV	chr16	2106721	C	T	497	260	237	0.476	TSC2	ENSG00000103197	NON_SYNONYMOUS_CODING	ENST00000350773.4:c.725C>T	ENSP00000344383.4:p.Thr242Ile	8/41	-	-	-
S06	SNV	chr16	2106721	C	T	497	260	237	0.476	TSC2	ENSG00000103197	NON_SYNONYMOUS_CODING	ENST00000439673.2:c.614C>T	ENSP00000399232.2:p.Thr205Ile	7/39	-	-	-
S06	SNV	chr16	2106721	C	T	497	260	237	0.476	TSC2	ENSG00000103197	NON_SYNONYMOUS_CODING	ENST00000401874.2:c.725C>T	ENSP00000384468.2:p.Thr242Ile	8/40	-	-	-
S13	SNV	chr16	2110695	G	A	325	211	114	0.351	TSC2	ENSG00000103197	NON_SYNONYMOUS_CODING	ENST00000401874.2:c.1000G>A	ENSP00000384468.2:p.Val334Met	11/40	-	-	-
S13	SNV	chr16	2110695	G	A	325	211	114	0.351	TSC2	ENSG00000103197	NON_SYNONYMOUS_CODING	ENST00000219476.3:c.1000G>A	ENSP00000219476.3:p.Val334Met	11/42	YES	-	-
S13	SNV	chr16	2110695	G	A	325	211	114	0.351	TSC2	ENSG00000103197	NON_SYNONYMOUS_CODING	ENST00000382538.6:c.853G>A	ENSP00000371978.6:p.Val285Met	10/39	-	-	-
S13	SNV	chr16	2110695	G	A	325	211	114	0.351	TSC2	ENSG00000103197	NON_SYNONYMOUS_CODING	ENST00000350773.4:c.1000G>A	ENSP00000344383.4:p.Val334Met	11/41	-	-	-
S13	SNV	chr16	2110695	G	A	325	211	114	0.351	TSC2	ENSG00000103197	NON_SYNONYMOUS_CODING	ENST00000568454.1:c.1033G>A	ENSP00000454487.1:p.Val345Met	11/40	-	-	-
S13	SNV	chr16	2110695	G	A	325	211	114	0.351	TSC2	ENSG00000103197	NON_SYNONYMOUS_CODING	ENST00000439673.2:c.889G>A	ENSP00000399232.2:p.Val297Met	10/39	-	-	-
S13	SNV	chr16	2110695	G	A	325	211	114	0.351	TSC2	ENSG00000103197	NON_SYNONYMOUS_CODING	ENST00000353929.4:c.1000G>A	ENSP00000248099.4:p.Val334Met	11/41	-	-	-
S43	SNV	chr16	2126110	C	T	569	303	266	0.468	TSC2	ENSG00000103197	NON_SYNONYMOUS_CODING	ENST00000382538.6:c.2534C>T	ENSP00000371978.6:p.Ala845Val	23/39	-	-	-
S43	SNV	chr16	2126110	C	T	569	303	266	0.468	TSC2	ENSG00000103197	NON_SYNONYMOUS_CODING	ENST00000350773.4:c.2681C>T	ENSP00000344383.4:p.Ala894Val	24/41	-	-	-
S43	SNV	chr16	2126110	C	T	569	303	266	0.468	TSC2	ENSG00000103197	NON_SYNONYMOUS_CODING	ENST00000219476.3:c.2681C>T	ENSP00000219476.3:p.Ala894Val	24/42	YES	-	-
S43	SNV	chr16	2126110	C	T	569	303	266	0.468	TSC2	ENSG00000103197	NON_SYNONYMOUS_CODING	ENST00000568454.1:c.2714C>T	ENSP00000454487.1:p.Ala905Val	24/40	-	-	-
S43	SNV	chr16	2126110	C	T	569	303	266	0.468	TSC2	ENSG00000103197	NON_SYNONYMOUS_CODING	ENST00000439673.2:c.2570C>T	ENSP00000399232.2:p.Ala857Val	23/39	-	-	-
S43	SNV	chr16	2126110	C	T	569	303	266	0.468	TSC2	ENSG00000103197	NON_SYNONYMOUS_CODING	ENST00000353929.4:c.2681C>T	ENSP00000248099.4:p.Ala894Val	24/41	-	-	-
S43	SNV	chr16	2126110	C	T	569	303	266	0.468	TSC2	ENSG00000103197	NON_SYNONYMOUS_CODING	ENST00000401874.2:c.2681C>T	ENSP00000384468.2:p.Ala894Val	24/40	-	-	-
31	SNV	chr14	81558891	C	G	345	189	156	0.452	TSHR	ENSG00000165409	NON_SYNONYMOUS_CODING	ENST00000298171.2:c.484C>G	ENSP00000298171.2:p.Pro162Ala	6/10	-	0.0004	rs121908863,CM951243
31	SNV	chr14	81558891	C	G	345	189	156	0.452	TSHR	ENSG00000165409	NON_SYNONYMOUS_CODING	ENST00000342443.6:c.484C>G	ENSP00000340113.6:p.Pro162Ala	6/9	-	0.0004	rs121908863,CM951243
31	SNV	chr14	81558891	C	G	345	189	156	0.452	TSHR	ENSG00000165409	UTR_5_PRIME	ENST00000541158.2:c.484C>G	ENSP00000441235.2:p.Pro162Ala	7/11	YES	0.0004	rs121908863,CM951243
11	INDEL	chr21	44513110	G	+T	133	95	38	0.275	U2AF1	ENSG00000160201	UTR_3_PRIME	-	-	7/7	-	0.0002	rs556707314,rs568046085
11	INDEL	chr21	44513110	G	+T	133	95	38	0.275	U2AF1	ENSG00000160201	UTR_3_PRIME	-	-	8/8	YES	0.0002	rs556707314,rs568046085
11	INDEL	chr21	44513110	G	+T	133	95	38	0.275	U2AF1	ENSG00000160201	UTR_3_PRIME	-	-	8/8	-	0.0002	rs556707314,rs568046085
11	INDEL	chr21	44513110	G	+T	133	95	38	0.275	U2AF1	ENSG00000160201	UTR_3_PRIME	-	-	9/9	-	0.0002	rs556707314,rs568046085
62	SNV	chr21	44524453	C	A	175	110	65	0.371	U2AF1	ENSG00000160201	NON_SYNONYMOUS_CODING	ENST00000291552.4:c.104G>T	ENSP00000291552.4:p.Arg35Leu	2/8	YES	-	COSM211657
62	SNV	chr21	44524453	C	A	175	110	65	0.371	U2AF1	ENSG00000160201	UTR_5_PRIME	ENST00000398137.1:c.-183G>T	-	2/9	-	-	COSM211657
62	SNV	chr21	44524453	C	A	175	110	65	0.371	U2AF1	ENSG00000160201	UTR_5_PRIME	ENST00000459639.1:c.-116G>T	-	1/7	-	-	COSM211657
62	SNV	chr21	44524453	C	A	175	110	65	0.371	U2AF1	ENSG00000160201	NON_SYNONYMOUS_CODING	ENST00000380276.2:c.104G>T	ENSP00000369629.2:p.Arg35Leu	2/8	-	-	COSM211657
S23	SNV	chr21	44524456	G	A	593	304	289	0.487	U2AF1	ENSG00000160201	NON_SYNONYMOUS_CODING	ENST00000291552.4:c.101C>T	ENSP00000291552.4:p.Ser34Phe	2/8	YES	-	rs371769427,COSM166866,COSM146287,COSM1142948,COSM1190367
S23	SNV	chr21	44524456	G	A	593	304	289	0.487	U2AF1	ENSG00000160201	NON_SYNONYMOUS_CODING	ENST00000380276.2:c.101C>T	ENSP00000369629.2:p.Ser34Phe	2/8	-	-	rs371769427,COSM166866,COSM146287,COSM1142948,COSM1190367
S25	SNV	chr21	44524456	G	A	202	110	92	0.455	U2AF1	ENSG00000160201	NON_SYNONYMOUS_CODING	ENST00000291552.4:c.101C>T	ENSP00000291552.4:p.Ser34Phe	2/8	YES	-	rs371769427,COSM166866,COSM146287,COSM1142948,COSM1190367

S25	SNV	chr21	44524456	G	A	202	110	92	0.455	U2AF1	ENSG00000160201	NON_SYNONYMOUS_CODING	ENST00000380276.2:c.101C>T	ENSP00000369629.2:p.Ser34Phe	2/8	-	-	rs371769427,COSM166866,COSM146287,COSM1142948,COSM1190367
S26	SNV	chr21	44514777	T	G	316	185	131	0.415	U2AF1	ENSG00000160201	NON_SYNONYMOUS_CODING	ENST00000291552.4:c.470A>C	ENSP00000291552.4:p.Gln157Pro	6/8	YES	-	rs371246226,COSM211532,COSM211534,COSM1724986,COSM1318797
S26	SNV	chr21	44514777	T	G	316	185	131	0.415	U2AF1	ENSG00000160201	NON_SYNONYMOUS_CODING	ENST00000459639.1:c.251A>C	ENSP00000418705.1:p.Gln84Pro	5/7	-	-	rs371246226,COSM211532,COSM211534,COSM1724986,COSM1318797
S26	SNV	chr21	44514777	T	G	316	185	131	0.415	U2AF1	ENSG00000160201	NON_SYNONYMOUS_CODING	ENST00000398137.1:c.251A>C	ENSP00000381205.1:p.Gln84Pro	7/9	-	-	rs371246226,COSM211532,COSM211534,COSM1724986,COSM1318797
S26	SNV	chr21	44514777	T	G	316	185	131	0.415	U2AF1	ENSG00000160201	NON_SYNONYMOUS_CODING	ENST00000380276.2:c.470A>C	ENSP00000369629.2:p.Gln157Pro	6/8	-	-	rs371246226,COSM211532,COSM211534,COSM1724986,COSM1318797
S37	SNV	chr21	44524456	G	A	499	347	152	0.305	U2AF1	ENSG00000160201	NON_SYNONYMOUS_CODING	ENST00000291552.4:c.101C>T	ENSP00000291552.4:p.Ser34Phe	2/8	YES	-	rs371769427,COSM166866,COSM146287,COSM1142948,COSM1190367
S37	SNV	chr21	44524456	G	A	499	347	152	0.305	U2AF1	ENSG00000160201	NON_SYNONYMOUS_CODING	ENST00000380276.2:c.101C>T	ENSP00000369629.2:p.Ser34Phe	2/8	-	-	rs371769427,COSM166866,COSM146287,COSM1142948,COSM1190367
37	INDEL	chr11	118267187	C	-CA	163	93	70	0.429	UBE4A	ENSG00000110344	NON_SYNONYMOUS_CODING	ENST00000431736.2:c.1036G>C	ENSP00000252108.3:p.Val346Leu	8/20	-	-	-
37	INDEL	chr11	118267187	C	-CA	163	93	70	0.429	UBE4A	ENSG00000110344	UTR_3_PRIME	ENST00000252108.3:c.*33delC	-	20/20	-	-	-
37	INDEL	chr11	118267187	C	-CA	163	93	70	0.429	UBE4A	ENSG00000110344	UTR_3_PRIME	ENST00000543354.1:c.*33delC	-	11/11	-	-	-
37	SNV	chr11	118244320	G	C	108	59	49	0.45	UBE4A	ENSG00000110344	NON_SYNONYMOUS_CODING	ENST00000431736.2:c.1057G>C	ENSP00000387362.2:p.Val353Leu	8/20	YES	-	-
40	SNV	chr17	5051908	T	C	145	83	62	0.428	USP6	ENSG00000129204	NON_SYNONYMOUS_CODING	ENST00000304328.5:c.1536T>C	ENSP00000305473.5:p.Leu513Pro	23/31	-	-	-
40	SNV	chr17	5051908	T	C	145	83	62	0.428	USP6	ENSG00000129204	NON_SYNONYMOUS_CODING	ENST00000250066.6:c.2489T>C	ENSP00000250066.6:p.Leu830Pro	22/30	-	-	-
40	SNV	chr17	5051908	T	C	145	83	62	0.428	USP6	ENSG00000129204	UTR_3_PRIME	ENST00000332776.4:c.*16T>C	-	29/37	-	-	-
S20	SNV	chr17	5037221	C	T	1260	726	534	0.424	USP6	ENSG00000129204	NON_SYNONYMOUS_CODING	ENST00000332776.4:c.424C>T	ENSP00000328010.4:p.His142Tyr	15/37	-	-	-
S20	SNV	chr17	5037221	C	T	1260	726	534	0.424	USP6	ENSG00000129204	NON_SYNONYMOUS_CODING	ENST00000250066.6:c.424C>T	ENSP00000250066.6:p.His142Tyr	7/30	-	-	-
S20	SNV	chr17	5037221	C	T	1260	726	534	0.424	USP6	ENSG00000129204	NON_SYNONYMOUS_CODING	ENST00000574788.1:c.424C>T	ENSP00000460380.1:p.His142Tyr	15/38	YES	-	-
S21	SNV	chr19	6822232	C	T	86	49	37	0.43	VAV1	ENSG00000141968	SPLICE_SITE_REGION	ENST00000602142.1:c.450C>T	ENST00000602142.1:c.450C>T(p.%3D)	5/27	YES	-	-
S21	SNV	chr19	6822232	C	T	86	49	37	0.43	VAV1	ENSG00000141968	SPLICE_SITE_REGION	ENST00000596764.1:c.450C>T	ENST00000596764.1:c.450C>T(p.%3D)	5/26	-	-	-
S21	SNV	chr19	6822232	C	T	86	49	37	0.43	VAV1	ENSG00000141968	SPLICE_SITE_REGION	ENST00000599806.1:c.285C>T	ENST00000599806.1:c.285C>T(p.%3D)	5/27	-	-	-
S21	SNV	chr19	6822232	C	T	86	49	37	0.43	VAV1	ENSG00000141968	SPLICE_SITE_REGION	ENST00000539284.1:c.255C>T	ENST00000539284.1:c.255C>T(p.%3D)	5/26	-	-	-
S21	SNV	chr19	6822232	C	T	86	49	37	0.43	VAV1	ENSG00000141968	SPLICE_SITE_REGION	ENST00000304076.2:c.450C>T	ENST00000304076.2:c.450C>T(p.%3D)	5/26	-	-	-
1	SNV	chr1	68564225	T	G	207	106	101	0.488	WLS	ENSG00000116729	UTR_3_PRIME	ENST00000540432.1:c.*90A>C	-	12/12	-	-	-
1	SNV	chr1	68564225	T	G	207	106	101	0.488	WLS	ENSG00000116729	UTR_3_PRIME	ENST00000354777.2:c.*90A>C	-	12/12	YES	-	-
34	SNV	chr1	68591776	C	T	166	88	78	0.47	WLS	ENSG00000116729	UTR_3_PRIME	ENST00000370976.3:c.*133G>A	-	11/11	-	-	-
34	SNV	chr1	68591776	C	T	166	88	78	0.47	WLS	ENSG00000116729	UTR_3_PRIME	ENST00000262348.4:c.*133G>A	-	12/12	-	-	-
19	SNV	chr11	324 13569	A	G	388	214	174	0.448	WT1	ENSG00000184937	NON_SYNONYMOUS_CODING	ENST00000379079.2:c.745T>C	ENSP00000368370.2:p.Ser249Pro	9/10	-	-	-
19	SNV	chr11	324 13569	A	G	388	214	174	0.448	WT1	ENSG00000184937	NON_SYNONYMOUS_CODING	ENST00000452863.3:c.1330T>C	ENSP00000415516.3:p.Ser444Pro	8/9	-	-	-
19	SNV	chr11	324 13569	A	G	388	214	174	0.448	WT1	ENSG00000184937	NON_SYNONYMOUS_CODING	ENST00000332351.3:c.1381T>C	ENSP00000331327.3:p.Ser461Pro	9/10	YES	-	-
19	SNV	chr11	324 13569	A	G	388	214	174	0.448	WT1	ENSG00000184937	NON_SYNONYMOUS_CODING	ENST00000448076.3:c.1381T>C	ENSP00000413452.3:p.Ser461Pro	9/10	-	-	-
19	SNV	chr11	324 13569	A	G	388	214	174	0.448	WT1	ENSG00000184937	NON_SYNONYMOUS_CODING	ENST00000530998.1:c.694T>C	ENSP00000435307.1:p.Ser232Pro	8/9	-	-	-
S21	SNV	chr11	324 13544	T	G	271	180	91	0.336	WT1	ENSG00000184937	NON_SYNONYMOUS_CODING	ENST00000452863.3:c.1355A>C	ENSP00000415516.3:p.His452Pro	8/9	-	-	CM061235
S21	INDEL	chr11	324 13542	T	+CCGGGGGAGG	269	187	82	0.293	WT1	ENSG00000184937	FRAME_SHIFT	-	-	5/6	-	-	-
S21	SNV	chr11	324 13544	T	G	271	180	91	0.336	WT1	ENSG00000184937	NON_SYNONYMOUS_CODING	ENST00000332351.3:c.1406A>C	ENSP00000331327.3:p.His469Pro	9/10	YES	-	CM061235
S21	INDEL	chr11	324 17942	A	+G	329	183	146	0.442	WT1	ENSG00000184937	FRAME_SHIFT	-	-	6/9	-	0.0002	rs554416372,COSM49052,COSM1166757,COSM35983,COSM28946,COSM42121,COSM30694,COSM49042,COSM1166756
S21	SNV	chr11	324 13544	T	G	271	180	91	0.336	WT1	ENSG00000184937	NON_SYNONYMOUS_CODING	ENST00000527882.1:c.387A>C	ENSP00000435624.1:p.His130Pro	5/6	-	-	CM061235
S21	INDEL	chr11	324 13542	T	+CCGGGGGAGG	269	187	82	0.293	WT1	ENSG00000184937	FRAME_SHIFT	-	-	9/10	-	-	-
S21	INDEL	chr11	324 17942	A	+G	329	183	146	0.442	WT1	ENSG00000184937	FRAME_SHIFT	-	-	4/6	-	0.0002	rs554416372,COSM49052,COSM1166757,COSM35983,COSM28946,COSM42121,COSM30694,COSM49042,COSM1166756
S21	SNV	chr11	324 13544	T	G	271	180	91	0.336	WT1	ENSG00000184937	NON_SYNONYMOUS_CODING	ENST00000530998.1:c.719A>C	ENSP00000435307.1:p.His240Pro	8/9	-	-	CM061235
S21	SNV	chr11	324 13544	T	G	271	180	91	0.336	WT1	ENSG00000184937	NON_SYNONYMOUS_CODING	ENST00000379079.2:c.770A>C	ENSP00000368370.2:p.His257Pro	9/10	-	-	CM061235
S21	INDEL	chr11	324 17942	A	+G	329	183	146	0.442	WT1	ENSG00000184937	FRAME_SHIFT	-	-	7/10	-	0.0002	rs554416372,COSM49052,COSM1166757,COSM35983,COSM28946,COSM42121,COSM30694,COSM49042,COSM1166756
S21	SNV	chr11	324 13544	T	G	271	180	91	0.336	WT1	ENSG00000184937	NON_SYNONYMOUS_CODING	ENST00000448076.3:c.1406A>C	ENSP00000413452.3:p.His469Pro	9/10	-	-	CM061235
S21	INDEL	chr11	324 17942	A	+G	329	183	146	0.442	WT1	ENSG00000184937	FRAME_SHIFT	-	-	7/7	-	0.0002	rs554416372,COSM49052,COSM1166757,COSM35983,COSM28946,COSM42121,COSM30694,COSM49042,COSM1166756
S21	INDEL	chr11	324 17942	A	+G	329	183	146	0.442	WT1	ENSG00000184937	FRAME_SHIFT	-	-	7/10	YES	0.0002	rs554416372,COSM49052,COSM1166757,COSM35983,COSM28946,COSM42121,COSM30694,COSM49042,COSM1166756
S21	INDEL	chr11	324 13542	T	+CCGGGGGAGG	269	187	82	0.293	WT1	ENSG00000184937	FRAME_SHIFT	-	-	9/10	-	-	-
S21	INDEL	chr11	324 13542	T	+CCGGGGGAGG	269	187	82	0.293	WT1	ENSG00000184937	FRAME_SHIFT	-	-	8/9	-	-	-
S21	INDEL	chr11	324 17942	A	+G	329	183	146	0.442	WT1	ENSG00000184937	FRAME_SHIFT	-	-	6/9	-	0.0002	rs554416372,COSM49052,COSM1166757,COSM35983,COSM28946,COSM42121,COSM30694,COSM49042,COSM1166756
S21	INDEL	chr11	324 17942	A	+G	329	183	146	0.442	WT1	ENSG00000184937	FRAME_SHIFT	-	-	7/10	-	0.0002	rs554416372,COSM49052,COSM1166757,COSM35983,COSM28946,COSM42121,COSM30694,COSM49042,COSM1166756
S21	INDEL	chr11	324 13542	T	+CCGGGGGAGG	269	187	82	0.293	WT1	ENSG00000184937	FRAME_SHIFT	-	-	8/9	-	-	-
S21	INDEL	chr11	324 13542	T	+CCGGGGGAGG	269	187	82	0.293	WT1	ENSG00000184937	FRAME_SHIFT	-	-	9/10	YES	-	-
S44	INDEL	chr11	324 17909	C	+G	536	393	143	0.265	WT1	ENSG00000184937	FRAME_SHIFT	-	-	7/10	-	0.0000	rs142937387,COSM27307,COSM1317326,CM971595
S44	INDEL	chr11	324 17909	C	+G	536	393	143	0.265	WT1	ENSG00000184937	FRAME_SHIFT	-	-	7/10	-	0.0000	rs142937387,COSM27307,COSM1317326,CM971595
S44	INDEL	chr11	324 17909	C	+G	536	393	143	0.265	WT1	ENSG00000184937	FRAME_SHIFT	-	-	4/6	-	0.0000	rs142937387,COSM27307,COSM1317326,CM971595
S44	INDEL	chr11	324 17909	C	+G	536	393	143	0.265	WT1	ENSG00000184937	FRAME_SHIFT	-	-	7/10	YES	0.0000	rs142937387,COSM27307,COSM1317326,CM971595
S44	INDEL	chr11	324 17909	C	+G	536	393	143	0.265	WT1	ENSG00000184937	FRAME_SHIFT	-	-	6/9	-	0.0000	rs142937387,COSM27307,COSM1317326,CM971595
S44	INDEL	chr11	324 17909	C	+G	536	393	143	0.265	WT1	ENSG00000184937	FRAME_SHIFT	-	-	6/9	-	0.0000	rs142937387,COSM27307,COSM1317326,CM971595
1	SNV	chr3	14199767	T	C	400	214	186	0.465	XPC	ENSG00000154767	NON_SYNONYMOUS_CODING	ENST00000449060.2:c.1505A>G	ENSP00000404002.2:p.Glu502Gly	9/16	-	0.0002	rs563236303
1	SNV	chr3	14199767	T	C	400	214	186	0.465	XPC	ENSG00000154767	NON_SYNONYMOUS_CODING	ENST00000285021.7:c.1616A>G	ENSP00000285021.7:p.Glu539Gly	9/16	YES	0.0002	rs563236303
36	INDEL	chr2	61710181	T	-CTTCTTGTG	309	214	95	0.307	XPO1	ENSG00000082898	CODON_DELETION	ENST00000401558.2:c.2724delA	ENSP00000384863.2:p.Ala909LeufsTer22	22/25	YES	-	-
36	INDEL	chr2	61710181	T	-CTTCTTGTG	309	214	95	0.307	XPO1	ENSG00000082898	CODON_DELETION	ENST00000406957.1:c.2724delA	ENSP00000385559.1:p.Ala909LeufsTer22	23/26	-	-	-
36	INDEL	chr2	61710181	T	-CTTCTTGTG	309	214	95	0.307	XPO1	ENSG00000082898	CODON_DELETION	ENST00000404992.2:c.2724delA	ENSP00000385942.2:p.Ala909LeufsTer22	22/25	-	-	-
42	SNV	chr2	61761032	T	C	254	128	126	0.496	XPO1	ENSG00000082898	START_LOST	ENST00000420673.1:c.1A>G	ENSP00000393484.1:p.Met1?	1/3	-	-	-
42	SNV	chr2	61761032	T	C	254	128	126	0.496	XPO1	ENSG00000082898	START_LOST	ENST00000422552.1:c.1A>G	ENSP00000408190.1:p.Met1?	2/4	-	-	-
42	SNV	chr2	61761032	T	C	254	128	126	0.496	XPO1	ENSG00000082898	START_LOST	ENST00000436018.1:c.1A>G	ENSP00000390843.1:p.Met1?	2/3	-	-	-
42	SNV	chr2	61761032	T	C	254	128	126	0.496	XPO1	ENSG00000082898	START_LOST	ENST00000443240.1:c.1A>G	ENSP00000406428.1:p.Met1?	3/7	-	-	-
42	SNV	chr2	61761032	T	C	254	128	126	0.496	XPO1	ENSG00000082898	START_LOST	ENST00000401558.2:c.1A>G	ENSP00000384863.2:p.Met1?	2/25	YES	-	-
42	SNV	chr2	61761032	T	C	254	128	126	0.496	XPO1	ENSG00000082898	START_LOST	ENST00000449444.1:c.1A>G	ENSP00000406819.1:p.Met1?	2/3	-	-	-
42	SNV	chr2	61761032	T	C	254	128	126	0.496	XPO1	ENSG00000082898	START_LOST	ENST00000451765.1:c.1A>G	ENSP00000413853.1:p.Met1?	2/7	-	-	-
42	SNV	chr2	61761032	T	C	254	128	126	0.496	XPO1	ENSG00000082898	START_LOST	ENST00000457483.1:c.1A>G	ENSP00000394951.1:p.Met1?	3/5	-	-	-
42	SNV	chr2	61761032	T	C	254	128	126	0.496	XPO1	ENSG00000082898	START_LOST	ENST00000404992.2:c.1A>G	ENSP00000385942.2:p.Met1?	2/25	-	-	-
42	SNV	chr2	61761032	T	C	254	128	126	0.496	XPO1	ENSG00000082898	START_LOST	ENST00000406957.1:c.1A>G	ENSP00000385559.1:p.Met1?	3/26	-	-	-
S37	SNV	chr15	41106201	T	C	443	236	207	0.467	ZFYVE19	ENSG00000166140	NON_SYNONYMOUS_CODING	ENST00000336455.5:c.1240T>C	ENSP00000337824.5:p.Cys414Arg	11/12	-	-	-
S37	SNV	chr15	41106201	T	C	443	236	207	0.467	ZFYVE19	ENSG00000166140	NON_SYNONYMOUS_CODING	ENST00000355341.4:c.1270T>C	ENSP00000347498.4:p.Cys424Arg	10/11	YES	-	-
S37	SNV	chr15	41106201	T	C	443	236	207	0.467	ZFYVE19	ENSG00000166140	NON_SYNONYMOUS_CODING	ENST00000570108.1:c.1201T>C	ENSP00000456223.1:p.Cys401Arg	11/12	-	-	-
S37	SNV	chr15	41106201	T	C	443	236	207	0.467	ZFYVE19	ENSG00000166140	NON_SYNONYMOUS_CODING	ENST00000564258.1:c.745T>C	ENSP00000457617.1:p.Cys249Arg	10/11	-	-	-
S37	SNV	chr15	41106201	T	C	443	236	207	0.467	ZFYVE19	ENSG00000166140	NON_SYNONYMOUS_CODING	ENST00000299173.10:c.1066T>C	ENSP00000299173.10:p.Cys356Arg	9/10	-	-	-
10	SNV	chr16	89805472	A	C	411	245	166	0.404	ZNF276	ENSG00000158805	UTR_3_PRIME	ENST00000289816.5:c.*818A>C	-	11/11	-	-	rs553145380
48	SNV	chr16	89805472	A	C	490	266	224	0.457	ZNF276	ENSG00000158805	UTR_3_PRIME	ENST00000289816.5:c.*818A>C	-	11/11	-	-	rs553145380
30	SNV	chr12	6777244	G	A	176	88	88	0.5	ZNF384	ENSG00000126746	NON_SYNONYMOUS_CODING	ENST00000319770.3:c.1139C>T	ENSP00000321650.3:p.Pro380Leu	9/9	-	0.0002	rs52846551
30	SNV	chr12	6777244	G	A	176	88	88	0.5	ZNF384	ENSG00000126746	NON_SYNONYMOUS_CODING	ENST00000396795.1:c.1187C>T	ENSP00000380013.1:p.Pro396Leu	9/9	-	0.0002	rs52846551
30	SNV	chr12	6777244	G	A	176	88	88	0.5	ZNF384	ENSG00000126746	NON_SYNONYMOUS_CODING	ENST00000396801.3:c.1370C>T	ENSP00000380019.3:p.Pro457Leu	11/11	YES	0.0002	rs52846551
30	SNV	chr12	6777244	G	A	176	88	88	0.5	ZNF384	ENSG00000126746	NON_SYNONYMOUS_CODING	ENST00000396799.2:c.1187C>T	ENSP00000380017.2:p.Pro396Leu	10/10	-	0.0002	rs52846551
30	SNV	chr12	6777244	G	A	176	88	88	0.5	ZNF384	ENSG00000126746	NON_SYNONYMOUS_CODING	ENST00000355772.4:c.1022C>T	ENSP00000348018.4:p.Pro341Leu	9/9	-	0.0002	rs52846551
30	SNV	chr12	6777244	G	A	176	88	88	0.5	ZNF384	ENSG00000126746	NON_SYNONYMOUS_CODING	ENST00000361959.3:c.1370C>T	ENSP00000354592.3:p.Pro457Leu	11/11	-	0.0002	rs52846551
S61	SNV	chr18	22806969	T	C	924	510	414	0.448	ZNF521	ENSG00000198795	NON_SYNONYMOUS_CODING	ENST00000584787.1:c.2534A>G	ENSP00000463000.1:p.Ser845Gly	3/7	-	-	-
S61	SNV	chr18	22806969	T	C	924	510	414	0.448	ZNF521	ENSG00000198795	NON_SYNONYMOUS_CODING	ENST00000538137.2:c.9134A>G	ENSP00000440768.2:p.Ser3045Gly	4/8	-	-	-
S61	SNV	chr18	22806969	T	C	924	510	414	0.448	ZNF521	ENSG00000198795	NON_SYNONYMOUS_CODING	ENST00000631524.3:c.9134A>G	ENSP00000354794.3:p.Ser3045Gly	4/8	YES	-	-
10	SNV	chrX	15821819	T	G	49	15	34	0.68	ZRSR2	ENSG00000169249	STOP_GAINED	ENST00000307771.7:c.212T>G	ENSP00000303015.7:p.Leu71Ter	4/11	YES	-	-
S33	INDEL	chrX	15841230	C	+AGCCGG	136	104	32	0.235	ZRSR2	ENSG00000169249	CODON_CHANGE_PLUS_CODON_INSERTION	-	-	11/11	YES	-	-

APPENDIX C

Table of DMRs of hierarchical cluster of elderly (vs cluster of young)

DMR Nametag	chr	start	end	value	area	cluster	indexStart	indexEnd	L	clusterL	p.valueArea	fwer	Genes associated (distance, bp.)
HyperHier1	chr7	73752935	73753326	3.05	9.14	51887	45260	45262	3	3	3.29E-05	0.00E+00	GTF2IRD1 (-115169), CLIP2 (+49326)
HyperHier2	chr7	73820590	73820652	2.82	8.46	51890	45266	45268	3	3	5.01E-05	0.00E+00	GTF2IRD1 (-47679), CLIP2 (+116816)
HyperHier3	chr12	117042854	117042917	2.70	8.09	14911	71889	71891	3	3	6.22E-05	0.00E+00	MAP1LC3B2 (+45700), C12orf49 (+132980)
HyperHier4	chr17	75096202	75096382	2.53	7.58	26206	91773	91775	3	3	7.92E-05	0.00E+00	SEPT9 (-181200), SEC14L1 (+11461)
HyperHier5	chr11	133928292	133928346	2.45	7.35	12534	67633	67635	3	3	9.20E-05	0.00E+00	IGSF9B (-101439), JAM3 (-10501)
HyperHier6	chr15	100048371	100048500	2.44	7.31	20902	82531	82533	3	3	9.43E-05	0.00E+00	MEF2A (-57469), SYNM (+403150)
HyperHier7	chr5	177913434	177913485	2.41	7.22	46511	33899	33901	3	3	9.79E-05	0.00E+00	PHYKPL (-253674), COL23A1 (+104096)
HyperHier8	chr17	78560478	78560916	2.35	7.04	26472	92226	92228	3	3	1.08E-04	0.00E+00	CHMP6 (-404944), RPTOR (+41629)
HyperHier9	chr17	47296970	47297268	2.34	7.02	25425	90533	90535	3	3	1.10E-04	0.00E+00	ABI3 (+9530), PHOSPHO1 (+11009)
HyperHier10	chr17	1576449	1576724	2.32	6.97	23702	87286	87288	3	3	1.14E-04	0.00E+00	RILP (-23216), PRPF8 (+11544)
HyperHier11	chr4	3365280	3365442	2.32	11.58	41036	23745	23749	5	5	1.11E-05	0.00E+00	HGFAC (-78253), RGS12 (+49487)
HyperHier12	chr11	70253427	70253499	2.31	6.92	11007	65172	65174	3	3	1.17E-04	0.00E+00	CTTN (+8816), SHANK2 (+604909)
HyperHier13	chr7	151442351	151442481	2.28	6.85	53248	47688	47690	3	3	1.19E-04	0.00E+00	RHEB (-225406), PRKAG2 (+131794)
HyperHier14	chr13	114918456	114918702	2.24	6.73	17128	75992	75994	3	3	1.29E-04	0.00E+00	CDC16 (-81783), RASA3 (-20493)
HyperHier15	chr19	4543487	4544574	2.15	12.91	28000	95110	95115	6	6	5.91E-06	0.00E+00	LRG1 (-3545)
HyperHier16	chr11	70265971	70266172	2.15	6.44	11010	65179	65181	3	3	1.51E-04	0.00E+00	CTTN (+21425), SHANK2 (+592300)
HyperHier17	chr10	1505595	1505654	2.14	6.42	5978	55713	55715	3	3	1.53E-04	0.00E+00	IDI1 (-410515), ADARB2 (+274045)
HyperHier18	chr16	89023389	89023633	2.14	6.41	23434	86811	86813	3	3	1.55E-04	0.00E+00	PABPN1L (-90485), CBFA2T3 (+20101)
HyperHier19	chr11	1750302	1750939	2.12	10.62	9060	61431	61435	5	5	1.54E-05	0.00E+00	IFITM10 (+21200), KRTAP5-6 (+32196)
HyperHier20	chr11	69259247	69260271	2.12	16.94	10940	65010	65017	8	8	1.42E-06	0.00E+00	CCND1 (-196096), TPCN2 (+443394)
HyperHier21	chr16	88228257	88228776	2.11	6.34	23345	86661	86663	3	3	1.61E-04	0.00E+00	ZNF469 (-265362), BANP (+224893)
HyperHier22	chr16	89922218	89922539	2.09	6.27	23528	86977	86979	3	3	1.68E-04	0.00E+00	TCF25 (-17641), SPIRE2 (+27463)
HyperHier23	chr17	77901030	77901317	2.06	8.24	26423	92145	92148	4	4	5.79E-05	0.00E+00	CBX4 (-87946), TBC1D16 (+108473)
HyperHier24	chr6	158507753	158508188	2.06	8.23	49866	41339	41342	4	4	5.79E-05	0.00E+00	SERAC1 (+81331), SYNJ2 (+105052)
HyperHier25	chr3	66633255	66633408	2.06	6.17	38935	20146	20148	3	3	1.77E-04	0.00E+00	KBTBD8 (-415399), LRIG1 (-81976)
HyperHier26	chr2	240878313	240878541	2.04	6.13	34686	17577	17579	3	3	1.81E-04	0.00E+00	HDAC4 (-555784), NDUFA10 (+86392)
HyperHier27	chr16	87866696	87866833	2.04	6.12	23322	86624	86626	3	3	1.81E-04	0.00E+00	KLHDC4 (-67210), SLC7A5 (+36329)
HyperHier28	chr22	43166139	43166347	2.04	6.12	37343	104101	104103	3	3	1.82E-04	0.00E+00	A4GALT (-75251), ARFGAP3 (+87165)
HyperHier29	chr17	79484960	79485709	2.03	8.12	26613	92515	92518	4	4	6.10E-05	0.00E+00	FSCN2 (-10223), ACTG1 (-5528)
HyperHier30	chr1	10690443	10690588	2.03	6.08	830	1432	1434	3	3	1.86E-04	0.00E+00	PEX14 (+155572), CASZ1 (+166189)
HyperHier31	chr5	1555791	1555887	2.02	6.07	43502	28229	28231	3	3	1.87E-04	0.00E+00	LPCAT1 (-31747), MRPL36 (+244170)
HyperHier32	chr14	103410516	103411005	2.01	8.02	18778	78903	78906	4	4	6.50E-05	0.00E+00	AMN (+21768), CDC42BPB (+113038)
HyperHier33	chr20	19955436	19955868	1.98	7.91	35105	100026	100029	4	4	6.83E-05	0.00E+00	NAA20 (-42108), RIN2 (+88487)
HyperHier34	chr18	74114570	74114728	1.97	5.90	27366	94034	94036	3	3	2.08E-04	0.00E+00	SMIM21 (-974991), ZNF516 (+92497)
HyperHier35	chr12	1025529	1025755	1.95	5.86	12621	67774	67776	3	3	2.13E-04	0.00E+00	RAD52 (+33246), WNK1 (+163553)
HyperHier36	chr8	21767146	21767310	1.95	5.86	54204	49458	49460	3	3	2.13E-04	0.00E+00	GFRA2 (-120906), DOK2 (+4143)
HyperHier37	chr13	114065669	114066074	1.94	7.77	17068	75869	75872	4	4	7.26E-05	0.00E+00	GRTP1 (-47431), ADPRHL1 (+41967)
HyperHier38	chr17	26577563	26577877	1.94	5.81	24432	88633	88635	3	3	2.20E-04	0.00E+00	TMEM97 (-68401), NLK (+208538)
HyperHier39	chr18	60903834	60904418	1.93	7.72	27307	93911	93914	4	4	7.42E-05	0.00E+00	BCL2 (+83235), PHLPP1 (+521443)
HyperHier40	chr1	159046391	159047163	1.92	13.42	4100	7067	7073	7	7	4.97E-06	0.00E+00	AIM2 (-86)
HyperHier41	chr10	131589141	131589189	1.91	5.73	8509	60299	60301	3	3	2.32E-04	0.00E+00	EBF3 (+172940), MGMT (+323717)
HyperHier42	chr8	144260671	144260778	1.90	5.71	56208	52954	52956	3	3	2.36E-04	0.00E+00	GPIHBP1 (-34343), LY6H (-18597)
HyperHier43	chr11	113846918	113846937	1.90	5.69	11788	66461	66463	3	3	2.40E-04	0.00E+00	ZBTB16 (-83387), HTR3A (+1131)
HyperHier44	chr19	16466298	16466636	1.89	7.57	28607	96201	96204	4	4	8.02E-05	0.00E+00	KLF2 (+30839), EPS15L1 (+116356)
HyperHier45	chr17	1373605	1374051	1.89	5.67	23673	87225	87227	3	3	2.42E-04	0.00E+00	CRK (-14276), MYO1C (+22278)
HyperHier46	chr11	330536	331179	1.86	9.31	8861	61042	61046	5	5	2.98E-05	0.00E+00	B4GALNT4 (-38946), IFITM3 (-9808)
HyperHier47	chr10	1975562	1975631	1.84	5.52	6007	55770	55772	3	3	2.64E-04	0.00E+00	ADARB2 (-195927)
HyperHier48	chr11	628318	628677	1.83	7.30	8906	61133	61136	4	4	9.51E-05	0.00E+00	CDHR5 (-2420), SCT (-1355)
HyperHier49	chr16	85262708	85262920	1.82	5.47	23052	86140	86142	3	3	2.72E-04	0.00E+00	GSE1 (-384008), KIAA0513 (+165996)
HyperHier50	chr16	87441441	87441659	1.82	5.46	23279	86558	86560	3	3	2.77E-04	0.00E+00	MAP1LC3B (+16144), ZCCHC14 (+84101)
HyperHier51	chr10	3250774	3251149	1.82	5.46	6031	55808	55810	3	3	2.77E-04	0.00E+00	PITRM1 (-35959), KLF6 (+576505)
HyperHier52	chr5	1494980	1495356	1.82	9.08	43490	28211	28215	5	5	3.52E-05	0.00E+00	SLC6A3 (-49623), LPCAT1 (+28924)
HyperHier53	chr12	133294261	133294835	1.81	5.44	15619	73166	73168	3	3	2.80E-04	0.00E+00	PGAM5 (+7143), ANKLE2 (+43926)
HyperHier54	chr3	196351986	196352142	1.81	5.43	40761	23225	23227	3	3	2.81E-04	0.00E+00	NRROS (-14493), FBXO45 (+56505)
HyperHier55	chr17	79428036	79428750	1.81	7.23	26597	92481	92484	4	4	9.70E-05	0.00E+00	ACTG1 (+51414), ENSG00000171282 (+54853)
HyperHier56	chr17	3704471	3704621	1.80	12.58	23806	87474	87480	7	7	6.86E-06	0.00E+00	ITGAE (-9)
HyperHier57	chr1	152538380	152538888	1.79	7.18	3771	6485	6488	4	4	1.00E-04	0.00E+00	LCE3E (+614)

HyperHier58	chr11	94278324	94279068	1.79	17.94	11514	65988	65997	10	10	1.42E-06	0.00E+00	PIWIL4 (-21778), FUT4 (+1679)
HyperHier59	chr3	184297380	184297522	1.79	5.37	40501	22769	22771	3	3	2.93E-04	0.00E+00	VPS8 (-232480), EPHB3 (+17879)
HyperHier60	chr13	113646732	113646899	1.78	5.33	17016	75778	75780	3	3	2.98E-04	0.00E+00	F7 (-113305), MCF2L (+23281)
HyperHier61	chr3	48601117	48602040	1.75	8.74	38443	19247	19251	5	5	4.45E-05	0.00E+00	UCN2 (-373)
HyperHier62	chr17	58499679	58499911	1.74	12.20	25713	91004	91010	7	7	7.80E-06	0.00E+00	USP32 (-30300), APPBP2 (+103785)
HyperHier63	chr2	240196078	240197131	1.74	10.44	34654	17518	17523	6	6	1.73E-05	0.00E+00	HDAC4 (+126038), TWIST2 (+439932)
HyperHier64	chr19	2546844	2547067	1.74	5.22	27834	94845	94847	3	3	3.23E-04	0.00E+00	GADD45B (+70831), GNG7 (+155751)
HyperHier65	chr4	184320416	184320640	1.73	5.18	43128	27570	27572	3	3	3.30E-04	0.00E+00	CLDN24 (-76949), CDKN2AIP (-45216)
HyperHier66	chr11	67803951	67804112	1.73	5.18	10872	64895	64897	3	3	3.30E-04	0.00E+00	TCIRG1 (-2451)
HyperHier67	chr16	28943094	28943288	1.71	6.86	21963	84307	84310	4	4	1.19E-04	0.00E+00	CD19 (-69)
HyperHier68	chr6	32049053	32049829	1.71	23.99	47819	37236	37249	14	14	4.73E-07	0.00E+00	TNXB (-35536), ATF6B (+46576)
HyperHier69	chr2	3826610	3826980	1.71	8.55	30520	10362	10366	5	5	4.82E-05	0.00E+00	DCDC2C (+75342)
HyperHier70	chr10	75415704	75415875	1.71	6.84	7122	57713	57716	4	4	1.20E-04	0.00E+00	SYNPO2L (+40)
HyperHier71	chr7	2773782	2774195	1.71	6.84	50650	42815	42818	4	4	1.20E-04	0.00E+00	AMZ1 (+54833), GNA12 (+109969)
HyperHier72	chr19	45449099	45449301	1.71	5.13	29454	97765	97767	3	3	3.44E-04	0.00E+00	APOC2 (-1260)
HyperHier73	chr8	30413794	30413919	1.71	5.12	54499	50011	50013	3	3	3.47E-04	0.00E+00	SMIM18 (-82260), RBPMS (+171830)
HyperHier74	chr19	33764284	33764550	1.71	6.82	28956	96831	96834	4	4	1.21E-04	0.00E+00	SLC7A10 (-47661), CEBPA (+29053)
HyperHier75	chr16	89163343	89163800	1.71	6.82	23458	86865	86868	4	4	1.22E-04	0.00E+00	CDH15 (-74603), ACSF3 (+3355)
HyperHier76	chr16	85981310	85981947	1.70	10.22	23138	86292	86297	6	6	1.99E-05	0.00E+00	FOXF1 (-562504), IRF8 (+49220)
HyperHier77	chr10	468347	468412	1.70	5.09	5900	55574	55576	3	3	3.54E-04	0.00E+00	DIP2C (+267226), ZMYND11 (+287956)
HyperHier78	chr7	157342543	157342801	1.69	5.06	53471	48180	48182	3	3	3.61E-04	0.00E+00	DNAJB6 (+213012)
HyperHier79	chr12	54866945	54867542	1.68	11.79	13806	69887	69893	7	7	1.02E-05	0.00E+00	GTSF1 (-833)
HyperHier80	chr19	11289349	11289376	1.68	6.73	28349	95722	95725	4	4	1.28E-04	0.00E+00	SPC24 (-22879), KANK2 (+18880)
HyperHier81	chr4	3742095	3742357	1.67	6.70	41077	23827	23830	4	4	1.31E-04	0.00E+00	LRPAP1 (-207940), ADRA2C (-25899)
HyperHier82	chr8	1847999	1848143	1.67	5.02	53809	48747	48749	3	3	3.75E-04	0.00E+00	MYOM2 (-145084), ARHGEF10 (+75929)
HyperHier83	chr9	133768931	133769184	1.67	5.00	57538	54891	54893	3	3	3.78E-04	0.00E+00	QRFP (+167)
HyperHier84	chr7	158263204	158263277	1.67	5.00	53600	48405	48407	3	3	3.78E-04	0.00E+00	PTPRN2 (+117130)
HyperHier85	chr15	79296424	79296585	1.66	4.99	20403	81578	81580	3	3	3.81E-04	0.00E+00	CTSH (-59072), RASGRF1 (+86610)
HyperHier86	chr17	79004850	79006166	1.66	13.28	26518	92319	92326	8	8	5.20E-06	0.00E+00	BAIAP2 (-3454)
HyperHier87	chr5	176734633	176734858	1.66	4.97	46446	33788	33790	3	3	3.87E-04	0.00E+00	RAB24 (-4037), PRELID1 (+3971)
HyperHier88	chr17	1492087	1492206	1.65	4.96	23690	87261	87263	3	3	3.89E-04	0.00E+00	PITPNA (-26037), SLC43A2 (+39983)
HyperHier89	chr17	3600356	3600514	1.65	6.60	23803	87464	87467	4	4	1.37E-04	0.00E+00	P2RX5 (-737)
HyperHier90	chr5	36662804	36662950	1.65	4.94	44043	29215	29217	3	3	3.96E-04	0.00E+00	NIPBL (-213984), SLC1A3 (+56420)
HyperHier91	chr13	114261869	114261986	1.65	4.94	17085	75899	75901	3	3	3.97E-04	0.00E+00	TFDP1 (+22872), ATP4B (+50573)
HyperHier92	chr18	74691269	74691423	1.64	4.93	27383	94064	94066	3	3	3.97E-04	0.00E+00	MBP (+153379), ZNF236 (+155230)
HyperHier93	chr2	239008929	239009118	1.63	8.16	34594	17411	17415	5	5	6.08E-05	0.00E+00	FAM132B (-58625), SCLY (+39494)
HyperHier94	chr21	34774164	34775032	1.62	6.50	36284	102269	102272	4	4	1.45E-04	0.00E+00	IFNGR2 (-604)
HyperHier95	chr1	25291385	25292034	1.62	14.60	1562	2552	2560	9	13	2.60E-06	0.00E+00	RUNX3 (-209)
HyperHier96	chr6	312026	312105	1.62	4.87	46662	34178	34180	3	3	4.23E-04	0.00E+00	IRF4 (-79673), DUSP22 (+19969)
HyperHier97	chr9	139590572	139591234	1.62	6.48	57882	55344	55347	4	4	1.48E-04	0.00E+00	FAM69B (-16119), AGPAT2 (-9028)
HyperHier98	chr7	150548934	150549406	1.62	6.47	53158	47537	47540	4	4	1.48E-04	0.00E+00	AOC1 (+27455), KCNH2 (+126233)
HyperHier99	chr7	2563184	2563909	1.62	8.08	50612	42738	42742	5	5	6.22E-05	0.00E+00	LFNG (+4071), BRAT1 (+31814)
HyperHier100	chr16	46919021	46919194	1.61	6.46	22167	84631	84634	4	4	1.49E-04	0.00E+00	GPT2 (+818)
HyperHier101	chr5	1962311	1962554	1.61	4.84	43571	28402	28404	3	3	4.33E-04	0.00E+00	IRX4 (-75101), IRX2 (+789343)
HyperHier102	chr4	6927183	6927859	1.60	6.41	41247	24187	24190	4	4	1.54E-04	0.00E+00	TADA2B (-117597), TBC1D14 (+15949)
HyperHier103	chr1	24861604	24861919	1.60	8.01	1542	2522	2526	5	5	6.55E-05	0.00E+00	NCMAP (-20840), RCAN3 (+32375)
HyperHier104	chr6	169399683	169399760	1.60	4.80	50215	41951	41953	3	3	4.50E-04	0.00E+00	THBS2 (+254417), SMOC2 (+557891)
HyperHier105	chr7	150139870	150140172	1.60	4.80	53144	47503	47505	3	3	4.50E-04	0.00E+00	GIMAP8 (-7697), ZNF775 (+63597)
HyperHier106	chr5	1807695	1807759	1.60	4.79	43534	28288	28290	3	3	4.50E-04	0.00E+00	NDUFS6 (+6213), IRX4 (+79605)
HyperHier107	chr7	2764209	2764599	1.60	7.98	50646	42800	42804	5	5	6.60E-05	0.00E+00	AMZ1 (+45248), GNA12 (+119554)
HyperHier108	chr20	58630240	58630390	1.59	4.78	35874	101565	101567	3	3	4.53E-04	0.00E+00	CDH26 (+96833)
HyperHier109	chr20	60886007	60886158	1.59	4.77	35919	101629	101631	3	3	4.60E-04	0.00E+00	ADRM1 (+8022), LAMA5 (+56285)
HyperHier110	chr2	105372087	105372265	1.59	4.76	32326	13289	13291	3	3	4.61E-04	0.00E+00	POU3F3 (-99793)
HyperHier111	chr17	78190755	78190983	1.58	4.75	26450	92193	92195	3	3	4.69E-04	0.00E+00	SLC26A11 (-3360), SGSH (+3330)
HyperHier112	chr8	1616381	1616450	1.58	4.74	53781	48699	48701	3	3	4.72E-04	0.00E+00	CLN8 (-95512), DLGAP2 (+166884)
HyperHier113	chr4	3374483	3375144	1.58	7.88	41040	23757	23761	5	5	6.90E-05	0.00E+00	HGFAC (-68800), RGS12 (+58940)
HyperHier114	chr11	971527	972057	1.57	4.71	8955	61246	61248	3	3	4.79E-04	0.00E+00	AP2A2 (+45911), MUC6 (+64914)
HyperHier115	chr17	78753273	78754372	1.56	12.48	26481	92244	92251	8	8	6.86E-06	0.00E+00	CHMP6 (-211818), RPTOR (+234755)

HyperHier116	chr17	38717206	38717275	1.56	4.68	24900	89507	89509	3	3	4.95E-04	0.00E+00	TNS4 (-59392), CCR7 (+4483)
HyperHier117	chr7	553412	553693	1.56	4.67	50329	42140	42142	3	3	4.99E-04	0.00E+00	PDGFA (+5478), FAM20C (+360584)
HyperHier118	chr10	1401915	1402293	1.56	4.67	5970	55700	55702	3	3	5.00E-04	0.00E+00	IDI1 (-306994), ADARB2 (+377566)
HyperHier119	chr12	131417943	131418217	1.56	4.67	15411	72770	72772	3	3	5.00E-04	0.00E+00	GPR133 (-20372), RAN (+61656)
HyperHier120	chr1	1117473	1117625	1.55	4.65	72	142	144	3	3	5.06E-04	0.00E+00	TTLL10 (+8266), TNFRSF18 (+24402)
HyperHier121	chr13	112838022	112838611	1.55	9.29	16940	75626	75631	6	8	3.03E-05	0.00E+00	SPACA7 (-192316), SOX1 (+116404)
HyperHier122	chr17	79799358	79799549	1.54	7.72	26637	92565	92569	5	5	7.40E-05	0.00E+00	PPP1R27 (-6545), P4HB (+19116)
HyperHier123	chr7	4754502	4755415	1.54	10.79	50706	42907	42913	7	7	1.49E-05	0.00E+00	AP5Z1 (-60294), FOXK1 (+33019)
HyperHier124	chr1	92952641	92953279	1.54	10.78	3138	5381	5387	7	7	1.49E-05	0.00E+00	GFI1 (-3449)
HyperHier125	chr7	601503	601828	1.54	4.61	50337	42153	42155	3	3	5.25E-04	0.00E+00	PDGFA (-42635), PRKAR1B (+151178)
HyperHier126	chr16	88972776	88973054	1.53	4.60	23421	86782	86784	3	3	5.27E-04	0.00E+00	PABPN1L (-39889), CBFA2T3 (+70697)
HyperHier127	chr8	48091424	48091864	1.53	6.11	54778	50485	50488	4	4	1.83E-04	0.00E+00	SPIDR (-81523)
HyperHier128	chr3	195849011	195849674	1.52	6.09	40745	23189	23192	4	4	1.85E-04	0.00E+00	TFRC (-40283), ZDHHC19 (+88923)
HyperHier129	chr6	7468848	7468973	1.52	4.56	46904	34593	34595	3	3	5.42E-04	0.00E+00	DSP (-72897), RIOK1 (+79182)
HyperHier130	chr7	550508	550640	1.52	4.56	50328	42137	42139	3	3	5.46E-04	0.00E+00	PDGFA (+8457), FAM20C (+357605)
HyperHier131	chr16	89167018	89167481	1.51	6.06	23460	86872	86875	4	4	1.89E-04	0.00E+00	CDH15 (-70925), ACSF3 (+7033)
HyperHier132	chr14	103415458	103416268	1.51	6.05	18780	78909	78912	4	4	1.90E-04	0.00E+00	AMN (+26870), CDC42BPB (+107936)
HyperHier133	chr17	79425877	79426432	1.51	6.04	26596	92477	92480	4	4	1.92E-04	0.00E+00	ENSG00000171282 (+52615), ACTG1 (+53652)
HyperHier134	chr17	998432	998711	1.51	4.52	23637	87167	87169	3	3	5.60E-04	0.00E+00	ABR (+84596), TIMM22 (+98215)
HyperHier135	chr2	174889675	174890570	1.50	9.01	33372	15128	15133	6	6	3.69E-05	0.00E+00	SP3 (-59693), OLA1 (+223303)
HyperHier136	chr8	1860385	1861136	1.49	8.96	53810	48750	48755	6	6	3.83E-05	0.00E+00	MYOM2 (-132394), ARHGEF10 (+88619)
HyperHier137	chr4	1202509	1203168	1.49	19.41	40901	23483	23495	13	13	9.46E-07	0.00E+00	SPON2 (-36178), CTBP1 (+40086)
HyperHier138	chr19	49577162	49577303	1.49	4.47	29765	98308	98310	3	3	5.80E-04	0.00E+00	KCNA7 (-1035)
HyperHier139	chr9	140683212	140683797	1.49	5.96	58001	55518	55521	4	4	2.00E-04	0.00E+00	CACNA1B (-88736), EHMT1 (+170051)
HyperHier140	chr7	1545385	1545819	1.49	4.47	50510	42535	42537	3	3	5.84E-04	0.00E+00	INTS1 (-1599)
HyperHier141	chr6	31867757	31868144	1.49	8.93	47794	37138	37143	6	6	3.90E-05	0.00E+00	EHMT2 (-2490)
HyperHier142	chr17	78854232	78854418	1.49	4.46	26497	92281	92283	3	3	5.88E-04	0.00E+00	CHMP6 (-111316), RPTOR (+335257)
HyperHier143	chr16	474271	474528	1.49	5.94	21010	82732	82735	4	4	2.02E-04	0.00E+00	RAB11FIP3 (-1219)
HyperHier144	chr17	80346614	80346912	1.48	4.45	26720	92737	92739	3	3	5.92E-04	0.00E+00	HEXDC (-29431), UTS2R (+14610)
HyperHier145	chr6	157931791	157932180	1.48	8.90	49844	41303	41308	6	6	3.97E-05	0.00E+00	SNX9 (-312310), ZDHHC14 (+129821)
HyperHier146	chr6	1410167	1410497	1.48	7.41	46723	34291	34295	5	5	8.84E-05	0.00E+00	FOXC1 (-200349), FOXF2 (+20263)
HyperHier147	chr1	1600982	1601380	1.48	4.44	139	291	293	3	3	5.97E-04	0.00E+00	CDK11B (-12246), SLC35E2B (+22902)
HyperHier148	chr14	101908752	101909021	1.48	7.39	18712	78756	78760	5	5	9.03E-05	0.00E+00	DIO3 (-118801), ENSG00000269375 (+549622)
HyperHier149	chr17	81031536	81032067	1.48	4.43	26788	92860	92862	3	3	6.06E-04	0.00E+00	B3GNTL1 (-22116), METRNL (-5765)
HyperHier150	chr7	2802374	2802976	1.47	7.37	50653	42824	42828	5	5	9.10E-05	0.00E+00	GNA12 (+81283), AMZ1 (+83519)
HyperHier151	chr16	88111009	88111425	1.47	4.42	23339	86651	86653	3	3	6.08E-04	0.00E+00	ZNF469 (-382662), BANP (+107593)
HyperHier152	chr17	80581701	80581925	1.47	4.41	26738	92776	92778	3	3	6.13E-04	0.00E+00	WDR45B (+24598), FOXK2 (+104222)
HyperHier153	chr2	241585833	241585888	1.47	4.41	34731	17648	17650	3	3	6.13E-04	0.00E+00	AQP12B (+36456), GPR35 (+40999)
HyperHier154	chr14	105154557	105155327	1.47	10.29	18920	79168	79174	7	7	1.92E-05	0.00E+00	INF2 (-1032)
HyperHier155	chr7	119131	119499	1.47	4.40	50308	42111	42113	3	3	6.17E-04	0.00E+00	FAM20C (-73654)
HyperHier156	chr2	11917490	11917787	1.46	4.39	30745	10698	10700	3	3	6.25E-04	0.00E+00	TRIB2 (-939376), LPIN1 (+99918)
HyperHier157	chr16	4714229	4714815	1.46	8.78	21504	83643	83648	6	6	4.35E-05	0.00E+00	NUDT16L1 (-29173), MGRN1 (+39731)
HyperHier158	chr17	77806190	77807201	1.46	7.29	26414	92123	92127	5	5	9.53E-05	0.00E+00	CBX8 (-35781), CBX4 (+6532)
HyperHier159	chr2	209224225	209225123	1.46	7.29	33835	16084	16088	5	5	9.53E-05	0.00E+00	PTH2R (-46882), PIKFYVE (+93683)
HyperHier160	chr16	4103161	4103225	1.46	4.37	21464	83591	83593	3	3	6.37E-04	0.00E+00	CREBBP (-172466), ADCY9 (+62993)
HyperHier161	chr16	31091326	31091608	1.45	4.36	22067	84471	84473	3	3	6.40E-04	0.00E+00	ZNF646 (+5724), PRSS53 (+8817)
HyperHier162	chr11	1107580	1108026	1.45	4.36	8973	61280	61282	3	3	6.42E-04	0.00E+00	MUC5AC (-43777), MUC2 (+32928)
HyperHier163	chr6	155537901	155538055	1.45	8.72	49810	41251	41256	6	6	4.47E-05	0.00E+00	CLDN20 (-47169), TIAM2 (+384147)
HyperHier164	chr1	35250489	35250735	1.45	4.35	1955	3240	3242	3	4	6.48E-04	0.00E+00	GJA4 (-7987), GJB3 (+3822)
HyperHier165	chr6	167507447	167508226	1.44	11.55	50076	41710	41717	8	8	1.14E-05	0.00E+00	CCR6 (-28420), FGFR1OP (+95167)
HyperHier166	chr2	10637974	10638113	1.44	4.32	30688	10608	10610	3	3	6.69E-04	0.00E+00	ODC1 (-49414), NOL10 (+192049)
HyperHier167	chr7	157407820	157408632	1.44	7.19	53489	48218	48222	5	5	1.00E-04	0.00E+00	DNAJB6 (+278566), PTPRN2 (+972145)
HyperHier168	chr1	117529478	117530009	1.44	7.18	3516	6019	6023	5	5	1.00E-04	0.00E+00	CD101 (-14638), PTGFRN (+77065)
HyperHier169	chr10	131682386	131682509	1.43	4.29	8513	60308	60310	3	3	6.84E-04	0.00E+00	EBF3 (+79657), MGMT (+417000)
HyperHier170	chr12	132657317	132657669	1.43	5.71	15492	72893	72896	4	4	2.36E-04	0.00E+00	NOC4L (+28500), GALNT9 (+33080)
HyperHier171	chr16	85074868	85075249	1.43	7.13	23025	86099	86103	5	5	1.04E-04	0.00E+00	ZDHHC7 (-29951), KIAA0513 (-21759)
HyperHier172	chr22	17956453	17956641	1.43	7.13	36707	103021	103025	5	5	1.04E-04	0.00E+00	CECR2 (-80)
HyperHier173	chr8	42037170	42037527	1.43	4.28	54731	50408	50410	3	5	6.92E-04	0.00E+00	AP3M2 (+26885), PLAT (+27893)

HyperHier174	chr19	1998801	1999322	1.42	5.68	27776	94729	94732	4	4	2.41E-04	0.00E+00	BTBD2 (+16657), CSNK1G2 (+57874)
HyperHier175	chr12	53490352	53490632	1.42	4.26	13660	69489	69491	3	3	7.02E-04	0.00E+00	IGFBP6 (-737)
HyperHier176	chr2	11673928	11674557	1.42	4.26	30728	10668	10670	3	3	7.03E-04	0.00E+00	GREB1 (+1)
HyperHier177	chr11	66055170	66055683	1.42	5.67	10731	64637	64640	4	4	2.42E-04	0.00E+00	TMEM151A (-3914), YIF1A (+1214)
HyperHier178	chr8	6735472	6735559	1.42	4.25	53908	48925	48927	3	3	7.08E-04	0.00E+00	DEFB1 (+28)
HyperHier179	chr19	1168354	1169138	1.42	5.67	27649	94518	94521	4	4	2.43E-04	0.00E+00	SBNO2 (+5536), GPX4 (+64810)
HyperHier180	chr5	150020212	150020338	1.41	4.24	45856	32773	32775	3	3	7.14E-04	0.00E+00	MYOZ3 (-20169), SYNPO (+39633)
HyperHier181	chr3	52260349	52260671	1.41	5.65	38591	19534	19537	4	4	2.46E-04	0.00E+00	TLR9 (-331)
HyperHier182	chr16	3306524	3307037	1.41	5.64	21423	83515	83518	4	4	2.46E-04	0.00E+00	MEFV (-154)
HyperHier183	chr12	120688557	120688744	1.41	4.23	15017	72084	72086	3	3	7.23E-04	0.00E+00	RPLP0 (-49749), PXN (+14912)
HyperHier184	chr8	19539991	19540396	1.41	5.63	54173	49407	49410	4	5	2.49E-04	0.00E+00	INTS10 (-134457), CSGALNACT1 (-80188)
HyperHier185	chr10	72545053	72545473	1.41	4.22	7053	57606	57608	3	3	7.34E-04	0.00E+00	TBATA (-106)
HyperHier186	chr1	946875	947343	1.40	8.43	33	67	72	6	6	5.11E-05	0.00E+00	ISG15 (-1694)
HyperHier187	chr17	46648525	46648582	1.40	4.21	25341	90257	90259	3	3	7.41E-04	0.00E+00	HOXB3 (-17270), HOXB4 (+8919)
HyperHier188	chr12	133134938	133135463	1.40	5.60	15599	73118	73121	4	4	2.54E-04	0.00E+00	MUC8 (-84475), P2RX2 (-60202)
HyperHier189	chr16	85470485	85470674	1.40	4.20	23079	86184	86186	3	3	7.47E-04	0.00E+00	GSE1 (-176242), KIAA0513 (+373762)
HyperHier190	chr7	151504844	151505116	1.40	4.19	53251	47693	47695	3	3	7.48E-04	0.00E+00	RHEB (-287970), PRKAG2 (+69230)
HyperHier191	chr22	43525330	43525679	1.40	4.19	37353	104126	104128	3	3	7.49E-04	0.00E+00	MCAT (+13895), BIK (+18751)
HyperHier192	chr12	6745171	6745568	1.40	5.59	12849	68157	68160	4	5	2.56E-04	0.00E+00	LPAR5 (+243)
HyperHier193	chr16	71523395	71523912	1.40	8.38	22759	85685	85690	6	6	5.27E-05	0.00E+00	ENSG00000261611 (-440), ZNF19 (-413)
HyperHier194	chr11	67982542	67982703	1.40	4.19	10881	64916	64918	3	4	7.51E-04	0.00E+00	SUV420H1 (-1741)
HyperHier195	chr14	102394274	102394986	1.40	8.38	18739	78838	78843	6	6	5.30E-05	0.00E+00	DYNC1H1 (-36235), PPP2R5C (+166495)
HyperHier196	chr8	142913273	142913350	1.40	4.19	56106	52766	52768	3	3	7.53E-04	0.00E+00	PTP4A3 (+511219), TSNARE1 (+571231)
HyperHier197	chr16	88700622	88700818	1.39	4.17	23384	86717	86719	3	3	7.62E-04	0.00E+00	IL17C (-4281)
HyperHier198	chr7	158324928	158325118	1.39	4.17	53610	48422	48424	3	4	7.63E-04	0.00E+00	PTPRN2 (+55348)
HyperHier199	chr7	4850075	4850345	1.39	5.56	50717	42946	42949	4	4	2.60E-04	0.00E+00	AP5Z1 (+34957), PAPOLB (+51415)
HyperHier200	chr11	70440347	70440395	1.39	4.17	11022	65202	65204	3	3	7.66E-04	0.00E+00	CTTN (+195724), SHANK2 (+418001)
HyperHier201	chr16	88566483	88566748	1.39	4.17	23373	86699	86701	3	3	7.67E-04	0.00E+00	ZC3H18 (-70173), ZFPM1 (+46891)
HyperHier202	chr4	715865	716282	1.39	5.54	40842	23367	23370	4	4	2.63E-04	0.00E+00	PCGF3 (+16520), CPLX1 (+103912)
HyperHier203	chr2	239432223	239432329	1.38	4.15	34615	17455	17457	3	3	7.75E-04	0.00E+00	TWIST2 (-324397), ASB1 (+96811)
HyperHier204	chr22	29707524	29708028	1.38	5.53	36964	103452	103455	4	4	2.63E-04	0.00E+00	RASL10A (+3969), GAS2L1 (+4745)
HyperHier205	chr4	6890915	6891264	1.38	5.53	41242	24175	24178	4	4	2.64E-04	0.00E+00	TBC1D14 (-20482), KIAA0232 (+106623)
HyperHier206	chr6	170403557	170403962	1.38	4.14	50256	42017	42019	3	3	7.81E-04	0.00E+00	DLL1 (+195801), C6orf70 (+252039)
HyperHier207	chr16	86011615	86012468	1.38	6.90	23142	86302	86306	5	5	1.17E-04	0.00E+00	FOXF1 (-532091), IRF8 (+79633)
HyperHier208	chr13	114270609	114270794	1.38	4.13	17086	75902	75904	3	3	7.93E-04	0.00E+00	TFDP1 (+31646), ATP4B (+41799)
HyperHier209	chr14	101512592	101512653	1.37	4.10	18692	78723	78725	3	3	8.15E-04	0.00E+00	DIO3 (-515065), ENSG00000269375 (+153358)
HyperHier210	chr1	2437200	2438037	1.36	4.09	280	549	551	3	3	8.16E-04	0.00E+00	PANK4 (+20420), PLCH2 (+38721)
HyperHier211	chr5	964230	964654	1.36	4.09	43410	28064	28066	3	3	8.23E-04	0.00E+00	BRD9 (-71503), NKD2 (-44502)
HyperHier212	chr21	46929500	46929741	1.36	4.08	36631	102901	102903	3	3	8.25E-04	0.00E+00	SLC19A1 (+32764), COL18A1 (+54218)
HyperHier213	chr2	241807747	241808004	1.36	5.43	34743	17675	17678	4	4	2.81E-04	0.00E+00	AGXT (-20)
HyperHier214	chr1	15127680	15127851	1.36	4.07	1011	1712	1714	3	3	8.32E-04	0.00E+00	TMEM51 (-351262), KAZN (+202566)
HyperHier215	chr9	95820754	95820913	1.36	4.07	56923	54061	54063	3	3	8.34E-04	0.00E+00	SUSD3 (-155)
HyperHier216	chr7	2119340	2119608	1.36	4.07	50576	42671	42673	3	3	8.34E-04	0.00E+00	MAD1L1 (+153404), ELFN1 (+391719)
HyperHier217	chr11	59823993	59824161	1.36	5.42	10371	64040	64043	4	5	2.83E-04	0.00E+00	MS4A3 (-79)
HyperHier218	chr6	31560624	31560871	1.35	5.42	47741	36933	36936	4	4	2.84E-04	0.00E+00	NCR3 (+14)
HyperHier219	chr2	113884734	113885277	1.35	4.06	32479	13553	13555	3	3	8.39E-04	0.00E+00	PSD4 (-46542), IL1RN (+9532)
HyperHier220	chr17	79380400	79380706	1.35	6.77	26580	92446	92450	5	5	1.26E-04	0.00E+00	ENSG00000171282 (+7013), ACTG1 (+99254)
HyperHier221	chr2	42566404	42566556	1.35	4.06	31286	11555	11557	3	3	8.43E-04	0.00E+00	COX7A2L (+29670), EML4 (+169990)
HyperHier222	chr6	30859855	30860300	1.35	14.86	47663	36669	36679	11	11	2.60E-06	0.00E+00	GTF2H4 (-15883), DDR1 (+8217)
HyperHier223	chr17	57915665	57915773	1.35	5.40	25694	90965	90968	4	4	2.87E-04	0.00E+00	TUBD1 (+54577), VMP1 (+130893)
HyperHier224	chr1	25240865	25240938	1.35	4.05	1554	2538	2540	3	3	8.49E-04	0.00E+00	RUNX3 (+50599), CLIC4 (+169054)
HyperHier225	chr7	38356808	38357047	1.35	4.05	51331	44324	44326	3	3	8.51E-04	0.00E+00	STARD3NL (+139104), AMPH (+314239)
HyperHier226	chr13	114820966	114821832	1.35	5.39	17102	75936	75939	4	4	2.89E-04	0.00E+00	GAS6 (-254359), RASA3 (+76687)
HyperHier227	chr13	99135543	99135711	1.35	5.39	16593	74921	74924	4	4	2.90E-04	0.00E+00	RNF113B (-306108), STK24 (+38625)
HyperHier228	chr19	3811002	3811194	1.35	4.04	27933	94990	94992	3	3	8.59E-04	0.00E+00	MATK (-9288), ZFR2 (+57928)
HyperHier229	chr19	1155030	1155225	1.35	5.38	27644	94509	94512	4	4	2.90E-04	0.00E+00	SBNO2 (+19154), GPX4 (+51192)
HyperHier230	chr22	46472568	46473721	1.34	6.72	37470	104345	104349	5	5	1.29E-04	0.00E+00	WNT7B (-100136), PPARA (-73354)
HyperHier231	chr19	55549414	55549842	1.34	9.41	30139	99093	99099	7	7	2.81E-05	0.00E+00	GP6 (+4)

HyperHier232	chr8	1971042	1971471	1.34	4.01	53835	48793	48795	3	3	8.82E-04	0.00E+00	MYOM2 (-21898), ARHGEF10 (+199115)
HyperHier233	chr6	149805995	149806732	1.34	13.38	49698	41058	41067	10	10	5.20E-06	0.00E+00	ZC3H12D (-167)
HyperHier234	chr13	113975974	113976694	1.34	8.02	17054	75846	75851	6	6	6.50E-05	0.00E+00	LAMP1 (+24778), GRTP1 (+42107)
HyperHier235	chr7	2761640	2761760	1.33	4.00	50645	42797	42799	3	3	8.91E-04	0.00E+00	AMZ1 (+42544), GNA12 (+122258)
HyperHier236	chr20	62084549	62084833	1.33	4.00	36064	101881	101883	3	3	8.93E-04	0.00E+00	CHRNA4 (-91952), KCNQ2 (+19300)
HyperHier237	chr11	15136150	15136505	1.33	9.30	9537	62463	62469	7	7	2.98E-05	0.00E+00	INSC (+2358)
HyperHier238	chr21	44573854	44574022	1.33	3.99	36479	102663	102665	3	3	9.02E-04	0.00E+00	U2AF1 (-46241), CRYAA (-15180)
HyperHier239	chr2	220309434	220309687	1.33	3.98	34092	16524	16526	3	3	9.13E-04	0.00E+00	GMPPA (-54062), SPEG (+9993)
HyperHier240	chr7	1130697	1131133	1.32	5.30	50420	42321	42324	4	4	3.04E-04	0.00E+00	GPER (+4472), ZFAND2A (+68899)
HyperHier241	chr6	31088343	31088434	1.32	3.97	47693	36769	36771	3	3	9.17E-04	0.00E+00	CDSN (-166)
HyperHier242	chr3	11643368	11643630	1.32	3.97	37841	18187	18189	3	3	9.19E-04	0.00E+00	VGLL4 (+41899), ATG7 (+329397)
HyperHier243	chr5	79553267	79553606	1.32	3.97	44622	30338	30340	3	3	9.22E-04	0.00E+00	SERINC5 (-1539)
HyperHier244	chr19	13054427	13054718	1.32	5.27	28443	95921	95924	4	4	3.10E-04	0.00E+00	RAD23A (-2129)
HyperHier245	chr5	957511	957913	1.32	3.95	43407	28059	28061	3	3	9.36E-04	0.00E+00	BRD9 (-64773), NKD2 (-51232)
HyperHier246	chr11	3177622	3178024	1.32	5.26	9218	61892	61895	4	4	3.12E-04	0.00E+00	CARS (-99156), OSBPL5 (+8761)
HyperHier247	chr11	70477139	70477192	1.32	3.95	11027	65212	65214	3	3	9.39E-04	0.00E+00	CTTN (+232519), SHANK2 (+381206)
HyperHier248	chr10	102046685	102046768	1.31	3.94	7670	58666	58668	3	3	9.41E-04	0.00E+00	BLOC1S2 (-258)
HyperHier249	chr7	102553293	102553457	1.31	5.25	52420	46276	46279	4	4	3.15E-04	0.00E+00	LRRC17 (-63)
HyperHier250	chr17	39769012	39769213	1.31	3.94	24958	89593	89595	3	3	9.44E-04	0.00E+00	KRT16 (-108)
HyperHier251	chr5	368394	368804	1.31	3.94	43343	27953	27955	3	4	9.45E-04	0.00E+00	AHRR (+64308), C5orf55 (+74659)
HyperHier252	chr17	78058704	78058778	1.31	3.94	26442	92177	92179	3	3	9.47E-04	0.00E+00	GAA (-16762), CCDC40 (+48306)
HyperHier253	chr15	41805946	41806234	1.31	5.25	19529	80169	80172	4	4	3.17E-04	0.00E+00	LTK (-86)
HyperHier254	chr17	46651186	46652938	1.31	18.36	25343	90261	90274	14	14	9.46E-07	0.00E+00	HOXB3 (-20778), HOXB4 (+5411)
HyperHier255	chr10	21796152	21796441	1.31	3.93	6336	56312	56314	3	3	9.56E-04	0.00E+00	NEBL (-609766), SKIDA1 (+18314)
HyperHier256	chr17	78755379	78755442	1.31	3.93	26482	92252	92254	3	3	9.58E-04	0.00E+00	CHMP6 (-210230), RPTOR (+236343)
HyperHier257	chr2	8685837	8686087	1.31	3.93	30614	10499	10501	3	3	9.58E-04	0.00E+00	ID2 (-133013)
HyperHier258	chr10	134399880	134400506	1.31	5.23	8683	60637	60640	4	4	3.21E-04	0.00E+00	INPP5A (+48869), NKX6-2 (+199363)
HyperHier259	chr13	114917199	114917586	1.31	6.53	17127	75987	75991	5	5	1.42E-04	0.00E+00	CDC16 (-82969), RASA3 (-19307)
HyperHier260	chr3	122043799	122044172	1.31	3.92	39458	20898	20900	3	3	9.65E-04	0.00E+00	CSTA (-105)
HyperHier261	chr17	79297435	79297618	1.30	3.91	26558	92403	92405	3	3	9.75E-04	0.00E+00	SLC38A10 (-28422), TMEM105 (+6947)
HyperHier262	chr6	170268691	170268808	1.30	3.89	50247	42002	42004	3	3	9.91E-04	0.00E+00	C6orf70 (+117029), DLL1 (+330811)
HyperHier263	chr1	15392793	15392907	1.30	3.89	1024	1729	1731	3	3	9.92E-04	0.00E+00	TMEM51 (-86178), KAZN (+467650)
HyperHier264	chr13	114829170	114830313	1.30	9.07	17106	75945	75951	7	7	3.57E-05	0.00E+00	GAS6 (-262702), RASA3 (+68344)
HyperHier265	chr3	46718941	46719086	1.30	3.89	38389	19144	19146	3	3	9.97E-04	0.00E+00	ALS2CL (+16177), TDGF1 (+100287)
HyperHier266	chr6	32905114	32905320	1.29	5.17	47923	37650	37653	4	9	3.35E-04	0.00E+00	HLA-DMB (+3630), PSMB9 (+83279)
HyperHier267	chr17	76227255	76227422	1.29	5.17	26283	91907	91910	4	5	3.35E-04	0.00E+00	TMEM235 (-645)
HyperHier268	chr4	3287376	3287777	1.29	3.87	41033	23740	23742	3	3	1.01E-03	0.00E+00	RGS12 (-28297), HTT (+211169)
HyperHier269	chr2	86038423	86038802	1.29	5.15	32070	12887	12890	4	4	3.38E-04	0.00E+00	ATOH8 (+57596), ST3GAL5 (+77524)
HyperHier270	chr19	427022	427263	1.28	3.84	27520	94308	94310	3	3	1.04E-03	0.00E+00	THEG (-51130), SHC2 (+33853)
HyperHier271	chr1	1052600	1053027	1.28	6.38	57	108	112	5	5	1.56E-04	0.00E+00	C1orf159 (-1336)
HyperHier272	chr10	1531243	1531529	1.28	5.10	5984	55729	55732	4	4	3.52E-04	0.00E+00	IDI1 (-436276), ADARB2 (+248284)
HyperHier273	chr8	124215033	124215374	1.27	5.09	55771	52284	52287	4	4	3.53E-04	0.00E+00	ZHX1 (+71531), TBC1D31 (+130284)
HyperHier274	chr1	92946700	92947961	1.27	7.61	3137	5375	5380	6	6	7.90E-05	0.00E+00	GLMN (-182798), GFI1 (+2180)
HyperHier275	chr11	1469746	1470155	1.26	3.79	9025	61377	61379	3	3	1.08E-03	0.00E+00	BRSK2 (+37569), MOB2 (+38025)
HyperHier276	chr20	35233380	35233582	1.26	3.78	35377	100580	100582	3	3	1.09E-03	0.00E+00	C20orf24 (-744)
HyperHier277	chr10	126222996	126223401	1.26	3.78	8357	60038	60040	3	3	1.09E-03	0.00E+00	LHPP (+72795), FAM53B (+209420)
HyperHier278	chr17	37331531	37331768	1.26	5.03	24801	89298	89301	4	4	3.72E-04	0.00E+00	ARL5C (-8332), CACNB1 (+22306)
HyperHier279	chr15	100890907	100891257	1.25	5.02	20925	82565	82568	4	4	3.75E-04	0.00E+00	ADAMTS17 (-8872), CERS3 (+193843)
HyperHier280	chr13	47472064	47472360	1.25	11.26	16174	74185	74193	9	9	1.23E-05	0.00E+00	HTR2A (-2039)
HyperHier281	chr2	233981788	233981999	1.25	3.75	34382	17062	17064	3	3	1.13E-03	0.00E+00	ATG16L1 (-178323), INPP5D (+56705)
HyperHier282	chr7	157224017	157224041	1.25	3.75	53455	48157	48159	3	3	1.13E-03	0.00E+00	DNAJB6 (+94369)
HyperHier283	chr17	80196719	80197538	1.25	6.24	26690	92663	92667	5	5	1.72E-04	0.00E+00	SLC16A3 (+5566), CSNK1D (+34478)
HyperHier284	chr3	13663398	13663538	1.24	3.73	37910	18320	18322	3	3	1.15E-03	0.00E+00	FBLN2 (+72837), WNT7A (+258150)
HyperHier285	chr18	72166016	72166324	1.24	3.73	27355	94008	94010	3	3	1.15E-03	0.00E+00	CNDP1 (-35505), CNDP2 (+2727)
HyperHier286	chr17	7960092	7960424	1.24	3.73	24039	87967	87969	3	3	1.15E-03	0.00E+00	ALOX15B (+17923), ALOX12B (+30763)
HyperHier287	chr11	2308362	2308589	1.23	4.94	9150	61710	61713	4	4	3.95E-04	0.00E+00	ASCL2 (-16294), C11orf21 (+14667)
HyperHier288	chr7	4752951	4753002	1.23	3.70	50705	42904	42906	3	3	1.18E-03	0.00E+00	AP5Z1 (-62276), FOXK1 (+31037)
HyperHier289	chr12	54954337	54954400	1.23	3.70	13811	69900	69902	3	3	1.18E-03	0.00E+00	PDE1B (+11235), PPP1R1A (+28074)

HyperHier290	chr7	148801711	148802155	1.23	3.70	53091	47409	47411	3	3	1.19E-03	0.00E+00	ZNF786 (-14136), ZNF425 (+21505)
HyperHier291	chr17	1521312	1521483	1.23	3.70	23695	87270	87272	3	3	1.19E-03	0.00E+00	PITPNA (-55288), SLC43A2 (+10732)
HyperHier292	chr1	45097282	45098125	1.23	6.16	2357	3902	3906	5	5	1.78E-04	0.00E+00	TMEM53 (+42523), RNF220 (+226838)
HyperHier293	chr17	40715222	40715281	1.23	6.15	25031	89706	89710	5	5	1.79E-04	0.00E+00	MLX (-3838)
HyperHier294	chr11	70455140	70455304	1.23	3.69	11025	65208	65210	3	3	1.21E-03	0.00E+00	CTTN (+210575), SHANK2 (+403150)
HyperHier295	chr19	54566838	54567279	1.23	3.69	30084	99000	99002	3	3	1.21E-03	0.00E+00	VSTM1 (+148)
HyperHier296	chr6	32054441	32055046	1.23	12.27	47824	37261	37270	10	24	7.57E-06	0.00E+00	TNXB (-40839), ATF6B (+41273)
HyperHier297	chr6	31543169	31543686	1.23	14.72	47734	36897	36908	12	12	2.60E-06	0.00E+00	TNF (+84)
HyperHier298	chr2	159651559	159652019	1.23	4.90	33137	14704	14707	4	4	4.11E-04	0.00E+00	DAPL1 (-40)
HyperHier299	chr3	195946851	195947062	1.22	3.67	40752	23205	23207	3	3	1.22E-03	0.00E+00	SLC51A (+3582), PCYT1A (+67627)
HyperHier300	chr11	2206073	2206268	1.22	3.66	9139	61666	61668	3	3	1.24E-03	0.00E+00	TH (-13136), ASCL2 (+86011)
HyperHier301	chr18	24283375	24283696	1.22	4.88	27008	93304	93307	4	4	4.18E-04	0.00E+00	KCTD1 (-154137), AQP4 (+162246)
HyperHier302	chr1	3514921	3515194	1.22	3.66	437	821	823	3	3	1.24E-03	0.00E+00	MEGF6 (+13001), ARHGEF16 (+144068)
HyperHier303	chr18	10588980	10589360	1.22	6.09	26886	93055	93059	5	5	1.85E-04	0.00E+00	NAPG (+63139), PIEZO2 (+559417)
HyperHier304	chr4	1804007	1804492	1.22	3.65	40971	23641	23643	3	3	1.26E-03	0.00E+00	FGFR3 (+9211), LETM1 (+53724)
HyperHier305	chr8	144631768	144631915	1.22	4.86	56257	53042	53045	4	4	4.25E-04	0.00E+00	GSDMD (-3535)
HyperHier306	chr11	1769289	1769522	1.21	8.49	9063	61439	61445	7	7	5.01E-05	0.00E+00	IFITM10 (+2415), KRTAP5-6 (+50981)
HyperHier307	chr7	149543136	149543177	1.21	3.63	53122	47463	47465	3	3	1.28E-03	0.00E+00	ATP6V0E2 (-26900), ZNF862 (+7701)
HyperHier308	chr6	32026656	32027379	1.21	6.05	47810	37211	37215	5	5	1.91E-04	0.00E+00	TNXB (-13113), ATF6B (+68999)
HyperHier309	chr19	14591203	14591345	1.21	3.63	28530	96064	96066	3	3	1.29E-03	0.00E+00	PTGER1 (-5100), GIPC1 (+15670)
HyperHier310	chr11	2555406	2555576	1.21	3.63	9174	61755	61757	3	3	1.29E-03	0.00E+00	KCNQ1 (+89270), CDKN1C (+351620)
HyperHier311	chr11	70416238	70416309	1.21	3.62	11020	65198	65200	3	3	1.29E-03	0.00E+00	CTTN (+171627), SHANK2 (+442098)
HyperHier312	chr16	84840283	84840564	1.21	3.62	23011	86078	86080	3	3	1.30E-03	0.00E+00	CRISPLD2 (-13166), USP10 (+106840)
HyperHier313	chr5	169593	170081	1.21	3.62	43327	27920	27922	3	3	1.30E-03	0.00E+00	SDHA (-48519), PLEKHG4B (+29464)
HyperHier314	chr17	2886250	2886453	1.20	3.61	23774	87411	87413	3	3	1.30E-03	0.00E+00	OR1D5 (+80549), RAP1GAP2 (+186620)
HyperHier315	chr1	36615845	36616030	1.20	3.61	1994	3309	3311	3	3	1.30E-03	0.00E+00	TRAPPC3 (-868)
HyperHier316	chr17	6921295	6921465	1.20	3.61	23946	87748	87750	3	3	1.31E-03	0.00E+00	BCL6B (-4959)
HyperHier317	chr1	12655992	12656315	1.20	4.81	946	1613	1616	4	4	4.43E-04	0.00E+00	DHRS3 (+21583), TNFRSF1B (+429094)
HyperHier318	chr6	31544694	31544960	1.20	4.81	47735	36909	36912	4	4	4.44E-04	0.00E+00	TNF (+1483), LTB (+5375)
HyperHier319	chr8	144654887	144655447	1.20	6.01	56263	53056	53060	5	6	1.95E-04	0.00E+00	NAPRT1 (+5616), GSDMD (+19790)
HyperHier320	chr8	143279443	143279882	1.20	4.80	56132	52807	52810	4	4	4.47E-04	0.00E+00	TSNARE1 (+204880), PTP4A3 (+877570)
HyperHier321	chr2	242710824	242711046	1.20	3.60	34802	17782	17784	3	3	1.32E-03	0.00E+00	GAL3ST2 (-5305), D2HGDH (+36941)
HyperHier322	chr8	37731969	37732224	1.20	3.60	54617	50192	50194	3	3	1.32E-03	0.00E+00	BRF2 (-24675), RAB11FIP1 (+24875)
HyperHier323	chr13	107132340	107132458	1.20	3.60	16733	75220	75222	3	3	1.32E-03	0.00E+00	EFNB2 (+55063)
HyperHier324	chr3	127473609	127473936	1.20	3.59	39619	21149	21151	3	3	1.34E-03	0.00E+00	MGLL (+67928), ABTB1 (+81990)
HyperHier325	chr14	95837801	95837929	1.20	3.59	18486	78374	78376	3	3	1.34E-03	0.00E+00	CLMN (-51622), SYNE3 (+104308)
HyperHier326	chr10	134260981	134261655	1.19	4.77	8671	60615	60618	4	4	4.58E-04	0.00E+00	STK32C (-139874), INPP5A (-90006)
HyperHier327	chr1	1149192	1149661	1.19	4.77	79	156	159	4	4	4.60E-04	0.00E+00	TNFRSF4 (+85)
HyperHier328	chr17	77128445	77128623	1.19	3.56	26354	92013	92015	3	3	1.38E-03	1.00E-03	ENGASE (+57507), RBFOX3 (+350029)
HyperHier329	chr7	101361395	101361745	1.18	3.55	52381	46219	46221	3	3	1.39E-03	1.00E-03	CUX1 (-97721), MYL10 (-88994)
HyperHier330	chr18	74692116	74692235	1.18	3.55	27384	94067	94069	3	3	1.40E-03	1.00E-03	MBP (+152549), ZNF236 (+156060)
HyperHier331	chr19	51225848	51226849	1.18	8.27	29891	98590	98596	7	7	5.72E-05	0.00E+00	CLEC11A (-237)
HyperHier332	chr7	38350921	38351468	1.18	7.08	51329	44317	44322	6	6	1.07E-04	0.00E+00	STARD3NL (+133371), AMPH (+319972)
HyperHier333	chr12	27396706	27396937	1.18	4.72	13202	68711	68714	4	4	4.78E-04	0.00E+00	STK38L (-286)
HyperHier334	chr8	143694010	143694305	1.18	3.53	56164	52861	52863	3	3	1.41E-03	1.00E-03	ARC (+2675), BAI1 (+163367)
HyperHier335	chr1	3806715	3807312	1.18	3.53	472	913	915	3	3	1.41E-03	1.00E-03	C1orf174 (+9835), DFFB (+33169)
HyperHier336	chr19	4531638	4531961	1.18	4.71	27996	95091	95094	4	4	4.82E-04	0.00E+00	PLIN4 (-14084), PLIN5 (+3436)
HyperHier337	chr7	99970448	99971016	1.18	4.70	52275	46033	46036	4	4	4.85E-04	0.00E+00	PILRA (-336)
HyperHier338	chr11	1475775	1475899	1.18	3.53	9027	61383	61385	3	3	1.42E-03	1.00E-03	MOB2 (+32139), BRSK2 (+43455)
HyperHier339	chr22	19710880	19711327	1.17	8.22	36752	103105	103111	7	7	5.79E-05	0.00E+00	GP1BB (+636)
HyperHier340	chr8	144824625	144824658	1.17	4.69	56283	53107	53110	4	4	4.88E-04	0.00E+00	FAM83H (-8671), SCRIB (+72907)
HyperHier341	chr17	7107939	7108792	1.17	5.87	23963	87783	87787	5	5	2.13E-04	0.00E+00	ASGR1 (-25483), ACADVL (-12078)
HyperHier342	chr19	6482011	6482169	1.17	3.51	28110	95290	95292	3	3	1.45E-03	1.00E-03	DENND1C (-271)
HyperHier343	chr20	642436	642894	1.17	4.68	34859	99542	99545	4	4	4.93E-04	0.00E+00	SRXN1 (-8651), SCRT2 (+14158)
HyperHier344	chr6	52171973	52172083	1.17	3.51	48571	38892	38894	3	3	1.45E-03	1.00E-03	PAQR8 (-54898), MCM3 (-22393)
HyperHier345	chr10	31422942	31423550	1.17	3.51	6525	56693	56695	3	3	1.45E-03	1.00E-03	ZEB1 (-184855), ZNF438 (-102380)
HyperHier346	chr7	157451243	157451906	1.17	7.01	53494	48228	48233	6	6	1.11E-04	0.00E+00	DNAJB6 (+321915), PTPRN2 (+928796)
HyperHier347	chr6	6900945	6901065	1.17	3.50	46883	34566	34568	3	3	1.46E-03	1.00E-03	RREB1 (-207036), LY86 (+312664)

HyperHier348	chr7	2518188	2518824	1.17	4.67	50604	42723	42726	4	4	4.98E-04	0.00E+00	LFNG (-40970), CHST12 (+75283)
HyperHier349	chr1	1067099	1067223	1.17	3.50	59	116	118	3	3	1.46E-03	1.00E-03	TTLL10 (-42122), C1orf159 (-15683)
HyperHier350	chr17	75318655	75318882	1.17	3.50	26221	91801	91803	3	3	1.46E-03	1.00E-03	TNRC6C (-682368), SEPT9 (+41277)
HyperHier351	chr17	71361857	71361901	1.17	3.50	25995	91422	91424	3	3	1.47E-03	1.00E-03	CDC42EP4 (-53565), SDK2 (+278349)
HyperHier352	chr15	93580022	93580327	1.16	5.82	20749	82200	82204	5	5	2.17E-04	0.00E+00	RGMA (+36917), CHD2 (+137117)
HyperHier353	chr19	728040	728385	1.16	3.49	27575	94392	94394	3	3	1.48E-03	1.00E-03	MISP (-22913), PALM (+19112)
HyperHier354	chr6	32053100	32053748	1.16	6.98	47823	37255	37260	6	6	1.12E-04	0.00E+00	TNXB (-39519), ATF6B (+42593)
HyperHier355	chr6	30860866	30860905	1.16	3.48	47664	36680	36682	3	3	1.49E-03	1.00E-03	GTF2H4 (-15075), DDR1 (+9025)
HyperHier356	chr22	18042550	18042722	1.16	3.48	36708	103026	103028	3	3	1.49E-03	1.00E-03	SLC25A18 (-503)
HyperHier357	chr12	14413090	14413690	1.16	4.63	13056	68469	68472	4	4	5.15E-04	0.00E+00	GRIN2B (-280337), ATF7IP (-105226)
HyperHier358	chr6	170531180	170531847	1.16	5.79	50272	42045	42049	5	5	2.24E-04	0.00E+00	DLL1 (+68047), C6orf70 (+379793)
HyperHier359	chr6	41254433	41254885	1.16	4.63	48285	38392	38395	4	4	5.15E-04	0.00E+00	TREM1 (-202)
HyperHier360	chr1	246952118	246952508	1.16	5.78	5839	9964	9968	5	8	2.24E-04	0.00E+00	SCCPDH (+64964), AHCTF1 (+142413)
HyperHier361	chr12	133120548	133120760	1.16	3.47	15596	73112	73114	3	3	1.51E-03	1.00E-03	P2RX2 (-74749), MUC8 (-69928)
HyperHier362	chr8	42623718	42623946	1.16	4.62	54751	50439	50442	4	4	5.18E-04	0.00E+00	CHRNA6 (+97)
HyperHier363	chr20	44574271	44575021	1.16	4.62	35591	101029	101032	4	4	5.19E-04	0.00E+00	PCIF1 (+11379), ZNF335 (+26165)
HyperHier364	chr1	203155737	203156625	1.15	5.77	4798	8250	8254	5	5	2.25E-04	0.00E+00	CHI3L1 (-304)
HyperHier365	chr6	160241105	160241556	1.15	4.61	49905	41412	41415	4	4	5.22E-04	0.00E+00	MAS1 (-86643), PNLDC1 (+20030)
HyperHier366	chr17	43324322	43325791	1.15	12.69	25212	90033	90043	11	11	6.62E-06	0.00E+00	SPATA32 (+14422), FMNL1 (+25901)
HyperHier367	chr17	56355331	56355431	1.15	3.46	25668	90912	90914	3	3	1.53E-03	1.00E-03	MPO (+2915), LPO (+39594)
HyperHier368	chr10	3138418	3138534	1.15	3.45	6025	55799	55801	3	3	1.54E-03	1.00E-03	PFKP (+28764), PITRM1 (+76527)
HyperHier369	chr1	1851439	1852604	1.15	6.90	157	324	329	6	10	1.17E-04	0.00E+00	TMEM52 (-1310)
HyperHier370	chr2	74208590	74208655	1.15	3.44	31881	12550	12552	3	3	1.57E-03	1.00E-03	TET3 (-64827), DGUOK (+54670)
HyperHier371	chr1	26880547	26881009	1.14	5.72	1629	2691	2695	5	5	2.33E-04	0.00E+00	ARID1A (-141746), RPS6KA1 (+8435)
HyperHier372	chr17	78735268	78735596	1.14	5.72	26479	92236	92240	5	5	2.34E-04	0.00E+00	CHMP6 (-230209), RPTOR (+216364)
HyperHier373	chr4	1294566	1295078	1.14	5.72	40909	23513	23517	5	5	2.34E-04	0.00E+00	UVSSA (-46282), MAEA (+11183)
HyperHier374	chr12	25707564	25707667	1.14	3.43	13182	68687	68689	3	3	1.58E-03	1.00E-03	IFLTD1 (-1399)
HyperHier375	chr16	85433309	85433718	1.14	4.58	23074	86173	86176	4	4	5.38E-04	0.00E+00	GSE1 (-213308), KIAA0513 (+336696)
HyperHier376	chr3	9944183	9944537	1.14	3.43	37772	18057	18059	3	3	1.58E-03	1.00E-03	IL17RE (+57)
HyperHier377	chr16	28888747	28888925	1.14	3.42	21958	84300	84302	3	3	1.59E-03	1.00E-03	ATP2A1 (-890)
HyperHier378	chr6	35108605	35109298	1.14	6.84	48094	38123	38128	6	6	1.20E-04	0.00E+00	TCP11 (+472)
HyperHier379	chr19	35981347	35981549	1.14	4.56	29033	96993	96996	4	4	5.44E-04	0.00E+00	KRTDAP (-15)
HyperHier380	chr16	15596053	15596423	1.14	4.56	21721	83973	83976	4	4	5.44E-04	0.00E+00	MPV17L (+106602), KIAA0430 (+140785)
HyperHier381	chr10	126308381	126308552	1.14	3.41	8364	60048	60050	3	3	1.60E-03	1.00E-03	FAM53B (+124152), LHPP (+158063)
HyperHier382	chr19	50921745	50922484	1.14	6.82	29861	98513	98518	6	6	1.22E-04	0.00E+00	SPIB (-83), SPIB (-80)
HyperHier383	chr2	242702553	242702881	1.14	3.41	34801	17779	17781	3	3	1.61E-03	1.00E-03	GAL3ST2 (-13523), D2HGDH (+28723)
HyperHier384	chr2	25527266	25527366	1.13	3.40	30977	11089	11091	3	3	1.62E-03	1.00E-03	POMC (-135544), DNMT3A (+38143)
HyperHier385	chr4	681086	681440	1.13	3.40	40837	23355	23357	3	3	1.62E-03	1.00E-03	MFSD7 (+1967), MYL5 (+9552)
HyperHier386	chr10	375248	375830	1.13	3.40	5888	55555	55557	3	3	1.63E-03	1.00E-03	ZMYND11 (+195115), DIP2C (+360067)
HyperHier387	chr20	62168463	62168878	1.13	9.06	36086	101908	101915	8	8	3.62E-05	0.00E+00	PTK6 (+24)
HyperHier388	chr1	231155632	231156359	1.13	11.32	5554	9504	9513	10	10	1.21E-05	0.00E+00	TRIM67 (-142720), ARV1 (+41201)
HyperHier389	chr2	238499793	238500081	1.13	3.39	34566	17363	17365	3	3	1.64E-03	1.00E-03	RAB17 (-201)
HyperHier390	chr6	4890079	4890278	1.13	3.39	46841	34477	34479	3	3	1.64E-03	1.00E-03	CDYL (+113510), RPP40 (+114102)
HyperHier391	chr10	323260	323809	1.13	3.39	5885	55550	55552	3	3	1.64E-03	1.00E-03	ZMYND11 (+143111), DIP2C (+412071)
HyperHier392	chr2	3496821	3497214	1.13	3.38	30499	10322	10324	3	3	1.65E-03	1.00E-03	ADI1 (+26489), TRAPPC12 (+113561)
HyperHier393	chr12	58210661	58210878	1.12	3.37	13956	70140	70142	3	3	1.66E-03	1.00E-03	AVIL (-516)
HyperHier394	chr4	3516065	3516758	1.12	5.62	41059	23796	23800	5	5	2.51E-04	0.00E+00	LRPAP1 (+17874), DOK7 (+51374)
HyperHier395	chr12	110252616	110252831	1.12	3.37	14675	71332	71334	3	3	1.67E-03	1.00E-03	TRPV4 (-28)
HyperHier396	chr14	23623480	23624377	1.12	8.99	17262	76181	76188	8	8	3.74E-05	0.00E+00	CEBPE (-35104), SLC7A8 (+28920)
HyperHier397	chr9	35563534	35564052	1.12	3.36	56636	53686	53688	3	3	1.69E-03	1.00E-03	TESK1 (-41574), RUSC2 (+25164)
HyperHier398	chr2	98377791	98377939	1.12	3.36	32197	13089	13091	3	3	1.69E-03	1.00E-03	ZAP70 (+47842), TMEM131 (+234489)
HyperHier399	chr10	94448532	94448728	1.12	3.36	7493	58285	58287	3	3	1.69E-03	1.00E-03	HHEX (+685)
HyperHier400	chr6	167571324	167571630	1.12	3.36	50079	41728	41730	3	3	1.70E-03	1.00E-03	GPR31 (+340)
HyperHier401	chr12	132687617	132687729	1.12	3.36	15505	72923	72925	3	3	1.70E-03	1.00E-03	GALNT9 (+2900), NOC4L (+58680)
HyperHier402	chr6	166140451	166140706	1.12	3.36	50001	41573	41575	3	3	1.70E-03	1.00E-03	PDE10A (-65022), SDIM1 (+169146)
HyperHier403	chr17	32690127	32690569	1.12	4.48	24615	88945	88948	4	4	5.79E-04	0.00E+00	CCL1 (-98)
HyperHier404	chr20	47896448	47897451	1.12	7.81	35675	101174	101180	7	7	7.07E-05	0.00E+00	ZNFX1 (-2194)
HyperHier405	chr12	56323639	56323816	1.12	3.35	13841	69945	69947	3	3	1.72E-03	1.00E-03	WIBG (-2031), DGKA (-1028)

HyperHier406	chr10	11936672	11937255	1.11	3.34	6198	56103	56105	3	3	1.72E-03	1.00E-03	UPF2 (+140932), ECHDC3 (+152599)
HyperHier407	chr15	93591750	93591806	1.11	3.34	20750	82205	82207	3	3	1.73E-03	1.00E-03	RGMA (+25314), CHD2 (+148720)
HyperHier408	chr11	62573844	62574403	1.11	3.34	10502	64255	64257	3	3	1.73E-03	1.00E-03	NXF1 (-350)
HyperHier409	chr22	50968250	50968343	1.11	5.57	37643	104611	104615	5	5	2.59E-04	0.00E+00	SCO2 (-3429), TYMP (+147)
HyperHier410	chr17	79442037	79442206	1.11	3.33	26602	92500	92502	3	3	1.74E-03	1.00E-03	ACTG1 (+37685), ENSG00000171282 (+68582)
HyperHier411	chr20	62200091	62200854	1.11	7.78	36091	101925	101931	7	7	7.26E-05	0.00E+00	SRMS (-21616), HELZ2 (+3335)
HyperHier412	chr3	187453973	187454477	1.11	3.33	40574	22887	22889	3	3	1.74E-03	1.00E-03	RTP2 (-33880), BCL6 (+9290)
HyperHier413	chr17	80266884	80267030	1.11	3.33	26700	92687	92689	3	3	1.76E-03	1.00E-03	CSNK1D (-35350), CD7 (+8521)
HyperHier414	chr1	147737016	147737207	1.11	4.43	3620	6243	6246	4	4	6.03E-04	0.00E+00	NBPF24 (-137563), PPIAL4A (+218307)
HyperHier415	chr22	50173996	50174297	1.11	7.75	37571	104498	104504	7	7	7.35E-05	0.00E+00	BRD1 (+44305)
HyperHier416	chr17	943108	943342	1.11	3.32	23633	87160	87162	3	3	1.77E-03	1.00E-03	TIMM22 (+42868), ABR (+139943)
HyperHier417	chr10	45495677	45495971	1.10	4.40	6719	56992	56995	4	4	6.18E-04	0.00E+00	ZNF22 (-99), C10orf25 (+512)
HyperHier418	chr17	7082724	7083064	1.10	3.29	23962	87780	87782	3	3	1.82E-03	1.00E-03	ASGR1 (-11)
HyperHier419	chr8	143596671	143596947	1.10	4.38	56156	52850	52853	4	4	6.29E-04	0.00E+00	BAI1 (+66018), ARC (+100024)
HyperHier420	chr11	45115390	45115819	1.10	3.29	10082	63595	63597	3	3	1.83E-03	1.00E-03	TP53I11 (-143894), PRDM11 (-53289)
HyperHier421	chr4	3387281	3387341	1.10	3.29	41041	23762	23764	3	3	1.83E-03	1.00E-03	HGFAC (-56303), RGS12 (+71437)
HyperHier422	chr16	2082689	2083393	1.09	5.47	21296	83275	83279	5	5	2.73E-04	0.00E+00	TSC2 (-14425), SLC9A3R2 (+6172)
HyperHier423	chr17	46653615	46654330	1.09	4.37	25344	90275	90278	4	4	6.34E-04	0.00E+00	HOXB3 (-22689), HOXB4 (+3500)
HyperHier424	chr17	34207332	34207663	1.09	4.37	24675	89056	89059	4	4	6.37E-04	0.00E+00	CCL5 (+14)
HyperHier425	chr10	743286	743761	1.09	4.37	5933	55650	55653	4	4	6.37E-04	0.00E+00	DIP2C (-7918), LARP4B (+188178)
HyperHier426	chr17	79128918	79129078	1.09	4.37	26536	92356	92359	4	4	6.38E-04	0.00E+00	AATK (+10819), BAIAP2 (+120036)
HyperHier427	chr15	23810843	23811369	1.09	4.36	19054	79389	79392	4	9	6.39E-04	0.00E+00	MKRN3 (+652)
HyperHier428	chr21	45575559	45575832	1.09	3.27	36527	102737	102739	3	4	1.85E-03	1.00E-03	C21orf33 (+22209), ICOSLG (+85132)
HyperHier429	chr11	76858729	76859387	1.09	5.45	11269	65603	65607	5	5	2.78E-04	0.00E+00	MYO7A (+19748), GDPD4 (+139405)
HyperHier430	chr15	41914122	41914454	1.09	3.26	19534	80177	80179	3	3	1.89E-03	1.00E-03	MGA (-38322), TYRO3 (+63056)
HyperHier431	chr12	8068600	8068706	1.08	3.25	12917	68277	68279	3	3	1.89E-03	1.00E-03	SLC2A14 (-43233), SLC2A3 (+20218)
HyperHier432	chr11	2919689	2921176	1.08	13.01	9196	61847	61858	12	12	5.91E-06	0.00E+00	SLC22A18 (-3217)
HyperHier433	chr8	145008110	145008397	1.08	3.25	56301	53149	53151	3	3	1.90E-03	1.00E-03	EPPK1 (-55622), PLEC (+16790)
HyperHier434	chr20	35169380	35169886	1.08	7.57	35374	100570	100576	7	7	8.06E-05	0.00E+00	MYL9 (-254)
HyperHier435	chr10	22725309	22725587	1.08	3.23	6361	56378	56380	3	3	1.93E-03	1.00E-03	SPAG6 (+91049), PIP4K2A (+278036)
HyperHier436	chr12	57569768	57569940	1.08	4.31	13892	70019	70022	4	4	6.72E-04	0.00E+00	NXPH4 (-40724), LRP1 (+47572)
HyperHier437	chr7	100167198	100167564	1.08	3.23	52288	46050	46052	3	3	1.94E-03	1.00E-03	SAP25 (+3889), AGFG2 (+30533)
HyperHier438	chr7	20256996	20257178	1.08	3.23	50900	43360	43362	3	3	1.94E-03	2.00E-03	MACC1 (-74)
HyperHier439	chr12	123469284	123469669	1.08	3.23	15113	72251	72253	3	3	1.95E-03	2.00E-03	ABCB9 (-3281)
HyperHier440	chr1	27961563	27962037	1.07	7.52	1695	2815	2821	7	7	8.28E-05	0.00E+00	FGR (-12)
HyperHier441	chr2	3697378	3697542	1.07	3.22	30508	10340	10342	3	3	1.96E-03	2.00E-03	ALLC (-8325), COLEC11 (+47964)
HyperHier442	chr13	31309032	31309799	1.07	3.22	15934	73759	73761	3	3	1.96E-03	2.00E-03	ALOX5AP (-229)
HyperHier443	chr1	203144862	203145364	1.07	3.22	4795	8245	8247	3	3	1.96E-03	2.00E-03	MYBPH (-172)
HyperHier444	chr18	45458021	45458698	1.07	4.28	27153	93627	93630	4	4	6.91E-04	0.00E+00	SMAD2 (-848)
HyperHier445	chr6	44044325	44044507	1.07	3.21	48431	38638	38640	3	3	1.99E-03	2.00E-03	TMEM63B (-50235), VEGFA (+305972)
HyperHier446	chr11	68747554	68747636	1.07	3.20	10911	64957	64959	3	3	2.00E-03	2.00E-03	MRGPRD (+860)
HyperHier447	chr17	1686627	1686737	1.07	3.20	23714	87311	87313	3	3	2.00E-03	2.00E-03	SERPINF1 (+21429), SMYD4 (+46493)
HyperHier448	chr15	31515750	31515984	1.07	4.27	19296	79797	79800	4	9	6.99E-04	0.00E+00	KLF13 (-103191), TRPM1 (-62391)
HyperHier449	chr7	2059150	2060130	1.07	8.53	50568	42648	42655	8	9	4.89E-05	0.00E+00	MAD1L1 (+213238), ELFN1 (+331885)
HyperHier450	chr16	2133388	2133801	1.07	3.20	21304	83292	83294	3	3	2.01E-03	2.00E-03	NTHL1 (-35728), PKD1 (+52304)
HyperHier451	chr1	109204168	109204325	1.07	5.33	3280	5629	5633	5	5	2.98E-04	0.00E+00	HENMT1 (-99)
HyperHier452	chr4	1600072	1600531	1.06	3.19	40951	23609	23611	3	3	2.02E-03	2.00E-03	NKX1-1 (-200183), FAM53A (+85686)
HyperHier453	chr4	1221838	1222037	1.06	3.19	40904	23499	23501	3	3	2.03E-03	2.00E-03	SPON2 (-55277), CTBP1 (+20987)
HyperHier454	chr11	692963	693032	1.06	3.19	8912	61159	61161	3	3	2.03E-03	2.00E-03	TMEM80 (-2618), DEAF1 (+2534)
HyperHier455	chr1	32708234	32708485	1.06	4.25	1855	3054	3057	4	4	7.09E-04	0.00E+00	LCK (-8480), EIF3I (+20389)
HyperHier456	chr12	53297547	53298027	1.06	3.19	13651	69473	69475	3	3	2.03E-03	2.00E-03	KRT78 (-55009), KRT8 (+22466)
HyperHier457	chr19	1280841	1281113	1.06	3.18	27668	94554	94556	3	3	2.03E-03	2.00E-03	MUM1 (-4913)
HyperHier458	chr7	128579876	128579964	1.06	3.18	52782	46888	46890	3	3	2.04E-03	2.00E-03	IRF5 (+1926), TNPO3 (+115048)
HyperHier459	chr17	46641504	46642104	1.06	5.30	25338	90247	90251	5	5	3.01E-04	0.00E+00	HOXB3 (-10520), HOXB4 (+15669)
HyperHier460	chr13	44989900	44990174	1.06	3.18	16128	74110	74112	3	3	2.04E-03	2.00E-03	SERP2 (+42059), TSC22D1 (+160664)
HyperHier461	chr17	80190129	80190154	1.06	3.17	26687	92653	92655	3	3	2.06E-03	2.00E-03	SLC16A3 (-1421)
HyperHier462	chr11	67418045	67418365	1.06	11.61	10861	64866	64876	11	11	1.11E-05	0.00E+00	ACY3 (-75)
HyperHier463	chr2	24233923	24234017	1.06	3.17	30939	11011	11013	3	3	2.08E-03	2.00E-03	FKBP1B (-38614), MFSD2B (+1019)

HyperHier464	chr6	167536056	167536521	1.05	6.33	50077	41718	41723	6	6	1.62E-04	0.00E+00	CCR6 (+32)
HyperHier465	chr12	132663428	132663883	1.05	8.44	15495	72899	72906	8	8	5.06E-05	0.00E+00	GALNT9 (+26917), NOC4L (+34663)
HyperHier466	chr19	840737	841082	1.05	5.27	27593	94426	94430	5	6	3.09E-04	0.00E+00	PRTN3 (-53)
HyperHier467	chr6	31025933	31026287	1.05	4.21	47688	36756	36759	4	4	7.36E-04	0.00E+00	MUC22 (+47859), C6orf15 (+54226)
HyperHier468	chr20	62367372	62368256	1.05	9.48	36110	101959	101967	9	9	2.70E-05	0.00E+00	SLC2A4RG (-3400), LIME1 (-181)
HyperHier469	chr3	123813383	123813465	1.05	3.15	39512	20974	20976	3	5	2.11E-03	3.00E-03	KALRN (-104)
HyperHier470	chr2	84517474	84517950	1.05	8.41	32012	12793	12800	8	8	5.18E-05	0.00E+00	SUCLG1 (+168892)
HyperHier471	chr12	52585695	52586127	1.05	6.29	13602	69388	69393	6	6	1.65E-04	0.00E+00	KRT80 (-127)
HyperHier472	chr8	10489118	10489270	1.05	3.14	53982	49040	49042	3	3	2.14E-03	3.00E-03	RP1L1 (+23418), PRSS55 (+106138)
HyperHier473	chr6	44528765	44528865	1.05	3.14	48464	38677	38679	3	3	2.14E-03	3.00E-03	RUNX2 (-767291), CDC5L (+173553)
HyperHier474	chr8	144896176	144896307	1.04	3.13	56290	53126	53128	3	4	2.16E-03	3.00E-03	FAM83H (-80271), SCRIB (+1307)
HyperHier475	chr5	148810061	148810203	1.04	6.26	45806	32687	32692	6	6	1.69E-04	0.00E+00	IL17B (-51293), CSNK1A1 (+120395)
HyperHier476	chr8	133772742	133773115	1.04	3.13	55918	52473	52475	3	3	2.17E-03	3.00E-03	TMEM71 (-101)
HyperHier477	chr13	112616931	112617294	1.04	3.12	16896	75494	75496	3	3	2.18E-03	3.00E-03	SOX1 (-104800), TEX29 (+644098)
HyperHier478	chr6	792658	792778	1.04	4.16	46680	34205	34208	4	4	7.68E-04	1.00E-03	FOXQ1 (-519957), EXOC2 (-99607)
HyperHier479	chr1	156211409	156211896	1.04	10.39	3979	6839	6848	10	10	1.84E-05	0.00E+00	BGLAP (-100)
HyperHier480	chr12	322214	323242	1.04	12.45	12597	67727	67738	12	12	7.09E-06	0.00E+00	SLC6A12 (+961)
HyperHier481	chr10	1559112	1559249	1.04	3.11	5985	55733	55735	3	3	2.21E-03	3.00E-03	IDI1 (-464071), ADARB2 (+220489)
HyperHier482	chr17	80159399	80159595	1.04	3.11	26678	92639	92641	3	3	2.21E-03	3.00E-03	FASN (-103289), SLC16A3 (-32066)
HyperHier483	chr17	76875678	76876239	1.04	4.14	26322	91958	91961	4	4	7.80E-04	1.00E-03	USP36 (-38990), ENSG00000178404 (+23324)
HyperHier484	chr2	64834106	64834430	1.04	3.11	31614	12127	12129	3	3	2.21E-03	3.00E-03	SERTAD2 (+46779), AFTPH (+82803)
HyperHier485	chr17	79432709	79433150	1.03	4.14	26600	92494	92497	4	4	7.86E-04	1.00E-03	ACTG1 (+46877), ENSG00000171282 (+59390)
HyperHier486	chr5	66461884	66462662	1.03	4.13	44374	29833	29836	4	4	7.90E-04	1.00E-03	CD180 (+30354), MAST4 (+570084)
HyperHier487	chr2	103352296	103352711	1.03	3.10	32308	13262	13264	3	3	2.23E-03	3.00E-03	MFSD9 (+809)
HyperHier488	chr12	133000497	133000830	1.03	3.10	15568	73068	73070	3	5	2.24E-03	4.00E-03	GALNT9 (-310091), MUC8 (+50062)
HyperHier489	chr14	95786621	95786801	1.03	3.09	18481	78363	78365	3	3	2.24E-03	4.00E-03	CLMN (-468)
HyperHier490	chr5	1503595	1503979	1.03	3.09	43491	28216	28218	3	3	2.26E-03	4.00E-03	SLC6A3 (-58242), LPCAT1 (+20305)
HyperHier491	chr6	6588737	6589075	1.03	4.12	46870	34548	34551	4	4	8.00E-04	1.00E-03	LY86 (+565)
HyperHier492	chr7	2653651	2654420	1.03	7.20	50626	42762	42768	7	7	9.96E-05	0.00E+00	TTYH3 (-17549), IQCE (+55404)
HyperHier493	chr5	176937350	176937527	1.03	4.11	46475	33839	33842	4	4	8.06E-04	1.00E-03	DOK3 (-581)
HyperHier494	chr19	55417436	55417647	1.03	4.11	30133	99084	99087	4	4	8.07E-04	1.00E-03	NCR1 (+34)
HyperHier495	chr16	4732262	4732406	1.03	3.08	21508	83654	83656	3	3	2.28E-03	6.00E-03	NUDT16L1 (-11361), MGRN1 (+57543)
HyperHier496	chr6	106583161	106583218	1.02	3.07	49084	39958	39960	3	3	2.30E-03	6.00E-03	PRDM1 (+48995), ATG5 (+190476)
HyperHier497	chr19	1854549	1854819	1.02	4.09	27762	94710	94713	4	4	8.17E-04	2.00E-03	REXO1 (-6232), KLF16 (+8883)
HyperHier498	chr6	31894990	31895598	1.02	7.15	47797	37151	37157	7	7	1.02E-04	0.00E+00	ENSG00000244255 (-181), C2 (+40)
HyperHier499	chr12	108991777	108992148	1.02	4.09	14634	71254	71257	4	4	8.24E-04	3.00E-03	TMEM119 (-63)
HyperHier500	chr21	46341006	46341380	1.02	4.07	36583	102829	102832	4	5	8.31E-04	3.00E-03	PTTG1IP (-47441), ITGB2 (+7595)
HyperHier501	chr7	151547969	151548111	1.02	3.05	53254	47698	47700	3	3	2.34E-03	7.00E-03	RHEB (-331030), PRKAG2 (+26170)
HyperHier502	chr12	112203795	112204327	1.02	3.05	14729	71413	71415	3	3	2.34E-03	7.00E-03	ALDH2 (-665)
HyperHier503	chr6	32043739	32044869	1.02	7.12	47816	37226	37232	7	7	1.05E-04	0.00E+00	TNXB (-30399), ATF6B (+51713)
HyperHier504	chr6	170418412	170418678	1.02	3.05	50259	42024	42026	3	3	2.34E-03	7.00E-03	DLL1 (+181016), C6orf70 (+266824)
HyperHier505	chr22	49765054	49765229	1.02	3.05	37551	104466	104468	3	3	2.34E-03	7.00E-03	BRD1 (+453310), FAM19A5 (+879870)
HyperHier506	chr2	105924245	105924659	1.02	3.05	32352	13342	13344	3	3	2.35E-03	7.00E-03	TGFBRAP1 (+22039), GPR45 (+66252)
HyperHier507	chr9	136566991	136567339	1.02	3.05	57668	55059	55061	3	4	2.35E-03	7.00E-03	SARDH (+37877), DBH (+65683)
HyperHier508	chr7	155276169	155276880	1.02	6.10	53336	47898	47903	6	10	1.84E-04	0.00E+00	RBM33 (-160848), EN2 (+25701)
HyperHier509	chr11	64627528	64627776	1.02	3.05	10625	64451	64453	3	3	2.35E-03	7.00E-03	CDC42BPG (-15611), EHD1 (+18539)
HyperHier510	chr19	35629603	35630474	1.02	9.14	29018	96953	96961	9	9	3.26E-05	0.00E+00	FXYD7 (-4115), LGI4 (-3935), FXYD1 (-887)
HyperHier511	chr1	153362927	153364020	1.02	6.09	3816	6552	6557	6	6	1.85E-04	0.00E+00	S100A8 (+190)
HyperHier512	chr6	32181764	32182013	1.01	3.04	47859	37498	37500	3	3	2.36E-03	7.00E-03	GPSM3 (-21206), NOTCH4 (+9955)
HyperHier513	chr17	74868604	74869001	1.01	3.04	26198	91759	91761	3	4	2.36E-03	7.00E-03	MGAT5B (+74)
HyperHier514	chr7	630473	630914	1.01	5.07	50340	42160	42164	5	5	3.60E-04	0.00E+00	PDGFA (-71663), PRKAR1B (+122150)
HyperHier515	chr11	1891872	1892608	1.01	11.14	9084	61485	61495	11	11	1.25E-05	0.00E+00	TNNT3 (-48552), LSP1 (+5845)
HyperHier516	chr5	143191565	143192067	1.01	4.04	45711	32511	32514	4	4	8.56E-04	4.00E-03	NR3C1 (-408562), YIPF5 (+358407)
HyperHier517	chr8	81599484	81599569	1.01	3.03	55265	51441	51443	3	3	2.39E-03	8.00E-03	ZNF704 (+187489), ZBTB10 (+201625)
HyperHier518	chr11	67350153	67350326	1.01	3.03	10852	64847	64849	3	3	2.39E-03	8.00E-03	GSTP1 (-826)
HyperHier519	chr12	6308758	6309025	1.01	3.03	12812	68084	68086	3	3	2.39E-03	8.00E-03	CD9 (+11)
HyperHier520	chr6	15504844	15506085	1.01	8.07	47090	34953	34960	8	8	6.24E-05	0.00E+00	DTNBP1 (+157808), JARID2 (+258938)
HyperHier521	chr10	135202522	135203200	1.01	7.06	8827	60964	60970	7	7	1.08E-04	0.00E+00	MTG1 (-4814), ENSG00000254536 (-1477)

HyperHier522	chr14	24801073	24801794	1.01	4.03	17309	76285	76288	4	6	8.65E-04	5.00E-03	ADCY4 (+2843), LTB4R (+18917)
HyperHier523	chr5	965717	966019	1.01	3.02	43411	28067	28069	3	3	2.42E-03	9.00E-03	BRD9 (-72929), NKD2 (-43076)
HyperHier524	chr8	145086659	145086882	1.00	3.01	56318	53196	53198	3	3	2.44E-03	1.10E-02	SPATC1 (+189)
HyperHier525	chr13	114880761	114881103	1.00	5.01	17118	75968	75972	5	5	3.77E-04	1.00E-03	GAS6 (-313892), RASA3 (+17154)
HyperHier526	chr20	62509164	62509309	1.00	3.01	36124	101989	101991	3	3	2.46E-03	1.10E-02	DNAJC5 (-17298), TPD52L2 (+12590)
HyperHier527	chr2	102608155	102608233	1.00	3.00	32283	13224	13226	3	3	2.47E-03	1.10E-02	IL1R2 (-112)
HyperHier528	chr14	23284518	23284968	1.00	3.00	17235	76133	76135	3	3	2.47E-03	1.10E-02	SLC7A7 (+269)
HyperHier529	chr8	141220177	141220490	1.00	3.00	56002	52602	52604	3	3	2.47E-03	1.10E-02	TRAPPC9 (+248344), C8orf17 (+276918)
HyperHier530	chr11	93466719	93467095	1.00	3.00	11494	65948	65950	3	3	2.48E-03	1.10E-02	C11orf54 (-7850), KIAA1731 (+72102)
HyperHier531	chr16	89573312	89573955	1.00	4.00	23501	86934	86937	4	4	8.92E-04	6.00E-03	SPG7 (-1177)
HyperHier532	chr6	161796785	161796854	1.00	3.00	49940	41473	41475	3	3	2.48E-03	1.10E-02	AGPAT4 (-101727)
HyperHier533	chr22	22901145	22902047	1.00	9.97	36839	103253	103262	10	11	2.22E-05	0.00E+00	PRAME (+100)
HyperHier534	chr11	64980819	64981297	1.00	3.99	10652	64490	64493	4	4	9.02E-04	6.00E-03	POLA2 (-48175), CAPN1 (+31125)
HyperHier535	chr10	90611782	90611911	1.00	2.99	7435	58186	58188	3	3	2.51E-03	1.20E-02	STAMBPL1 (-27644), LIPM (+49360)
HyperHier536	chr19	17918795	17919173	1.00	2.99	28689	96332	96334	3	3	2.51E-03	1.20E-02	B3GNT3 (+13065), INSL3 (+13399)
HyperHier537	chr12	107711489	107711648	1.00	2.99	14600	71202	71204	3	3	2.51E-03	1.20E-02	BTBD11 (-621)
HyperHier538	chr20	62318426	62318588	0.99	3.97	36105	101947	101950	4	4	9.16E-04	6.00E-03	TNFRSF6B (-9514), RTEL1-TNFRSF6B (+27751)
HyperHier539	chr19	50936038	50936172	0.99	3.97	29863	98525	98528	4	4	9.16E-04	6.00E-03	MYBPC2 (-55)
HyperHier540	chr20	20036653	20037150	0.99	4.96	35108	100032	100036	5	5	3.88E-04	1.00E-03	CRNKL1 (-212)
HyperHier541	chr7	999994	1000336	0.99	2.97	50384	42240	42242	3	3	2.55E-03	1.20E-02	ADAP1 (-5832), COX19 (+15070)
HyperHier542	chr11	43290958	43291043	0.99	2.97	10004	63415	63417	3	5	2.55E-03	1.20E-02	API5 (-42538)
HyperHier543	chr1	1118157	1118391	0.99	2.97	73	145	147	3	3	2.55E-03	1.20E-02	TTLL10 (+8991), TNFRSF18 (+23677)
HyperHier544	chr17	79282893	79283959	0.99	7.93	26557	92395	92402	8	8	6.74E-05	0.00E+00	SLC38A10 (-14321), TMEM105 (+21048)
HyperHier545	chr6	168106055	168106222	0.99	2.97	50098	41764	41766	3	4	2.56E-03	1.20E-02	TCP10 (-308185), C6orf123 (+91400)
HyperHier546	chr1	50574507	50574591	0.99	2.97	2504	4198	4200	3	4	2.56E-03	1.20E-02	ELAVL4 (+2585), DMRTA2 (+314623)
HyperHier547	chr1	203273571	203274134	0.99	2.97	4805	8264	8266	3	3	2.57E-03	1.20E-02	BTG2 (-811)
HyperHier548	chr6	167189213	167189543	0.99	2.96	50072	41704	41706	3	3	2.58E-03	1.20E-02	MPC1 (-392892), RPS6KA2 (+86661)
HyperHier549	chr7	99769432	99769602	0.99	2.96	52270	46020	46022	3	3	2.59E-03	1.20E-02	GAL3ST4 (-3144)
HyperHier550	chr19	11689715	11689997	0.99	6.90	28382	95788	95794	7	7	1.17E-04	0.00E+00	ACP5 (-62)
HyperHier551	chr10	45958759	45958885	0.99	2.96	6726	57002	57004	3	3	2.60E-03	1.20E-02	ALOX5 (+89147), MARCH8 (+131532)
HyperHier552	chr12	96389483	96390059	0.99	4.93	14404	70840	70844	5	5	3.99E-04	1.00E-03	HAL (+372)
HyperHier553	chr15	69754609	69754714	0.99	2.96	20103	81069	81071	3	3	2.60E-03	1.20E-02	RPLP1 (+9539), TLE3 (+635853)
HyperHier554	chr16	66612955	66613407	0.98	10.83	22578	85358	85368	11	11	1.49E-05	0.00E+00	CMTM2 (-179)
HyperHier555	chr11	67142001	67142268	0.98	2.95	10825	64795	64797	3	3	2.61E-03	1.20E-02	CLCF1 (-927)
HyperHier556	chr12	132854818	132855224	0.98	2.95	15522	72962	72964	3	6	2.61E-03	1.20E-02	GALNT9 (-164448), MUC8 (+195705)
HyperHier557	chr17	79419796	79420424	0.98	5.90	26592	92467	92472	6	6	2.08E-04	0.00E+00	ENSG00000171282 (+46570), ACTG1 (+59697)
HyperHier558	chr10	134211620	134211908	0.98	3.92	8664	60596	60599	4	4	9.59E-04	6.00E-03	INPP5A (-139560), STK32C (-90320)
HyperHier559	chr17	80398117	80398854	0.98	2.94	26725	92748	92750	3	3	2.65E-03	1.20E-02	OGFOD3 (-22034), C17orf62 (+10099)
HyperHier560	chr19	827429	828170	0.98	7.83	27588	94410	94417	8	8	7.02E-05	0.00E+00	AZU1 (-26)
HyperHier561	chr11	381709	381875	0.98	2.94	8872	61066	61068	3	3	2.66E-03	1.20E-02	PKP3 (-12425), B4GALNT4 (+11988)
HyperHier562	chr6	32076079	32076592	0.98	6.85	47834	37349	37355	7	8	1.20E-04	0.00E+00	TNXB (-62431), ATF6B (+19681)
HyperHier563	chr4	6695464	6695614	0.98	2.93	41232	24162	24164	3	3	2.67E-03	1.20E-02	S100P (+743)
HyperHier564	chr12	8834114	8834151	0.98	2.93	12932	68294	68296	3	3	2.67E-03	1.20E-02	MFAP5 (-18725), RIMKLB (-16385)
HyperHier565	chr16	85659946	85660534	0.98	2.93	23107	86235	86237	3	3	2.67E-03	1.20E-02	GSE1 (+13418), GINS2 (+62365)
HyperHier566	chr1	151030968	151031295	0.98	2.93	3698	6368	6370	3	3	2.67E-03	1.20E-02	MLLT11 (+898)
HyperHier567	chr20	43882990	43883746	0.98	5.86	35545	100929	100934	6	6	2.13E-04	0.00E+00	SLPI (-163)
HyperHier568	chr16	28518200	28518385	0.98	2.93	21946	84286	84288	3	3	2.68E-03	1.20E-02	IL27 (-138)
HyperHier569	chr2	240291068	240291657	0.98	3.90	34663	17539	17542	4	4	9.79E-04	6.00E-03	HDAC4 (+31280), TWIST2 (+534690)
HyperHier570	chr16	556039	556350	0.98	2.93	21014	82740	82742	3	3	2.69E-03	1.20E-02	CAPN15 (-21661), RAB11FIP3 (+80576)
HyperHier571	chr17	5137892	5138696	0.98	4.88	23896	87636	87640	5	5	4.19E-04	1.00E-03	SCIMP (-139)
HyperHier572	chr11	503807	504937	0.97	7.79	8890	61101	61108	8	8	7.14E-05	0.00E+00	RNH1 (-802)
HyperHier573	chr12	1609245	1609537	0.97	6.81	12626	67782	67788	7	7	1.22E-04	0.00E+00	FBXL14 (+93940), ERC1 (+508987)
HyperHier574	chr4	2943901	2944692	0.97	4.86	41018	23717	23721	5	5	4.24E-04	1.00E-03	MFSD10 (-8112), NOP14 (+20815)
HyperHier575	chr8	144416327	144416485	0.97	2.92	56229	52995	52997	3	3	2.72E-03	1.20E-02	TOP1MT (+660)
HyperHier576	chr6	33282885	33282971	0.97	3.89	47988	37928	37931	4	13	9.92E-04	6.00E-03	TAPBP (-939)
HyperHier577	chr17	75315837	75316784	0.97	5.83	26220	91795	91800	6	6	2.17E-04	0.00E+00	TNRC6C (-684826), SEPT9 (+38819)
HyperHier578	chr11	68779817	68780136	0.97	3.88	10919	64970	64973	4	4	9.97E-04	6.00E-03	MRGPRF (+900)
HyperHier579	chr12	124864528	124865130	0.97	4.86	15155	72310	72314	5	5	4.28E-04	1.00E-03	NCOR2 (+114969), ZNF664 (+407159)

HyperHier580	chr1	22141170	22141400	0.97	3.88	1418	2342	2345	4	4	9.98E-04	6.00E-03	LDLRAD2 (+2527), HSPG2 (+122505)
HyperHier581	chr12	52404134	52404422	0.97	3.88	13591	69374	69377	4	4	1.00E-03	6.00E-03	NR4A1 (-12338), GRASP (+3554)
HyperHier582	chr1	16163479	16164122	0.97	6.78	1056	1775	1781	7	7	1.25E-04	0.00E+00	SPEN (-10558), FBLIM1 (+72807)
HyperHier583	chr7	28998195	28998595	0.97	5.80	51133	44001	44006	6	14	2.21E-04	0.00E+00	CHN2 (-235633), CREB5 (+546251)
HyperHier584	chr11	102826565	102826680	0.97	2.90	11627	66165	66167	3	3	2.77E-03	1.20E-02	MMP13 (-160)
HyperHier585	chr7	129845574	129845832	0.97	2.90	52811	46931	46933	3	3	2.78E-03	1.20E-02	SSMEM1 (-1997), TMEM209 (-365)
HyperHier586	chr3	52099522	52099561	0.96	2.89	38584	19524	19526	3	3	2.79E-03	1.30E-02	DUSP7 (-8976), POC1A (+88886)
HyperHier587	chr17	8301118	8301192	0.96	2.89	24070	88017	88019	3	3	2.80E-03	1.30E-02	RNF222 (-11)
HyperHier588	chr1	203733971	203734559	0.96	5.78	4829	8295	8300	6	6	2.24E-04	0.00E+00	LAX1 (-39)
HyperHier589	chr17	61511683	61511829	0.96	2.89	25768	91111	91113	3	3	2.80E-03	1.30E-02	CYB561 (+11789), TANC2 (+424839)
HyperHier590	chr6	30130458	30132226	0.96	17.35	47589	36390	36407	18	19	1.42E-06	0.00E+00	TRIM10 (-2631), TRIM15 (+349)
HyperHier591	chr22	21986960	21987190	0.96	7.70	36820	103217	103224	8	8	7.50E-05	1.00E-03	YDJC (-2722)
HyperHier592	chr17	46645972	46646444	0.96	3.85	25339	90252	90255	4	4	1.03E-03	7.00E-03	HOXB3 (-14924), HOXB4 (+11265)
HyperHier593	chr6	64281439	64281846	0.96	6.73	48659	39031	39037	7	7	1.29E-04	1.00E-03	PTP4A1 (-274)
HyperHier594	chr17	78748077	78748494	0.96	2.87	26480	92241	92243	3	3	2.86E-03	1.50E-02	CHMP6 (-217355), RPTOR (+229218)
HyperHier595	chr19	14224992	14225172	0.96	3.83	28511	96031	96034	4	4	1.04E-03	7.00E-03	SAMD1 (-23562), PRKACA (+3474)
HyperHier596	chr22	50986511	50986962	0.96	2.87	37649	104627	104629	3	7	2.86E-03	1.50E-02	TYMP (-18293), SYCE3 (+14597)
HyperHier597	chr8	143085750	143086459	0.96	5.75	56116	52780	52785	6	6	2.31E-04	1.00E-03	TSNARE1 (+398438), PTP4A3 (+684012)
HyperHier598	chr1	95006322	95006729	0.96	3.83	3172	5430	5433	4	4	1.04E-03	7.00E-03	F3 (+830)
HyperHier599	chr11	71259341	71259581	0.96	2.87	11074	65309	65311	3	3	2.87E-03	1.50E-02	KRTAP5-9 (-5)
HyperHier600	chr6	32077744	32078624	0.96	8.61	47835	37356	37364	9	9	4.75E-05	1.00E-03	TNXB (-64279), ATF6B (+17833)
HyperHier601	chr19	42502731	42503412	0.95	4.77	29353	97599	97603	5	5	4.58E-04	2.00E-03	ATP1A3 (-4690)
HyperHier602	chr12	132670344	132671672	0.95	6.68	15497	72908	72914	7	7	1.32E-04	1.00E-03	GALNT9 (+19565), NOC4L (+42015)
HyperHier603	chr6	30698584	30698987	0.95	8.59	47648	36612	36620	9	9	4.75E-05	1.00E-03	TUBB (+10808), FLOT1 (+11724)
HyperHier604	chr10	88024529	88024673	0.95	5.72	7373	58073	58078	6	6	2.34E-04	1.00E-03	GRID1 (+101634)
HyperHier605	chr2	240153103	240153778	0.95	2.86	34648	17502	17504	3	5	2.90E-03	1.60E-02	HDAC4 (+169202), TWIST2 (+396768)
HyperHier606	chr12	6881590	6882083	0.95	5.71	12867	68184	68189	6	6	2.36E-04	1.00E-03	LAG3 (+151)
HyperHier607	chr16	87588559	87588893	0.95	2.85	23289	86574	86576	3	3	2.92E-03	1.60E-02	ZCCHC14 (-63075), JPH3 (-47785)
HyperHier608	chr2	219246468	219247055	0.95	3.80	33989	16307	16310	4	4	1.07E-03	7.00E-03	SLC11A1 (+10)
HyperHier609	chr1	109849854	109850562	0.95	3.80	3309	5668	5671	4	4	1.08E-03	7.00E-03	MYBPHL (-545)
HyperHier610	chr1	161171211	161171819	0.95	4.74	4180	7187	7191	5	5	4.70E-04	2.00E-03	ADAMTS4 (-2669), NDUFS2 (+2410)
HyperHier611	chr3	52812033	52812520	0.95	4.74	38612	19577	19581	5	5	4.70E-04	2.00E-03	ITIH1 (+669)
HyperHier612	chr13	114831142	114831705	0.95	2.85	17107	75952	75954	3	3	2.95E-03	1.60E-02	GAS6 (-264384), RASA3 (+66662)
HyperHier613	chr17	75315081	75315244	0.95	2.85	26219	91792	91794	3	3	2.95E-03	1.60E-02	TNRC6C (-685974), SEPT9 (+37671)
HyperHier614	chr2	174877357	174877566	0.95	2.84	33368	15122	15124	3	3	2.95E-03	1.60E-02	SP3 (-47032), OLA1 (+235964)
HyperHier615	chr13	111128054	111128102	0.95	2.84	16822	75371	75373	3	3	2.95E-03	1.60E-02	RAB20 (+86002), COL4A2 (+168464)
HyperHier616	chr6	90660913	90661242	0.95	3.79	48897	39491	39494	4	4	1.08E-03	7.00E-03	GJA10 (+56890), BACH2 (+345383)
HyperHier617	chr19	2302401	2303032	0.95	5.68	27803	94792	94797	6	6	2.41E-04	1.00E-03	LINGO3 (-10694), LSM7 (+25898)
HyperHier618	chr11	74178114	74178883	0.95	6.63	11176	65466	65472	7	7	1.36E-04	1.00E-03	KCNE3 (+174)
HyperHier619	chr11	68856817	68856906	0.95	2.84	10925	64986	64988	3	3	2.98E-03	1.60E-02	CCND1 (-598993), TPCN2 (+40497)
HyperHier620	chr16	1587684	1587842	0.94	2.83	21242	83184	83186	3	3	2.98E-03	1.60E-02	TMEM204 (+4189), IFT140 (+74348)
HyperHier621	chr20	31261784	31261994	0.94	2.83	35279	100420	100422	3	3	2.99E-03	1.60E-02	COMMD7 (+69914), ASXL1 (+315734)
HyperHier622	chr12	22590017	22590199	0.94	2.83	13142	68628	68630	3	3	2.99E-03	1.60E-02	ST8SIA1 (-102460), C2CD5 (+107372)
HyperHier623	chr17	78865263	78866235	0.94	6.61	26502	92291	92297	7	8	1.37E-04	1.00E-03	CHMP6 (-99892), RPTOR (+346681)
HyperHier624	chr2	241905733	241906357	0.94	4.71	34756	17700	17704	5	5	4.79E-04	2.00E-03	SNED1 (-32210), ENSG00000226321 (+44059)
HyperHier625	chr13	113707471	113707579	0.94	2.83	17034	75812	75814	3	3	3.01E-03	1.70E-02	F7 (-52596), MCF2L (+83990)
HyperHier626	chr7	157620445	157620943	0.94	3.77	53534	48300	48303	4	4	1.10E-03	7.00E-03	DNAJB6 (+491034), PTPRN2 (+759677)
HyperHier627	chr7	155584080	155584274	0.94	2.83	53359	47946	47948	3	3	3.01E-03	1.70E-02	SHH (+20790), RBM33 (+146804)
HyperHier628	chr8	145051491	145051795	0.94	2.82	56313	53188	53190	3	3	3.02E-03	1.70E-02	PLEC (-26599), PARP10 (+8979)
HyperHier629	chr8	21916635	21916853	0.94	3.76	54223	49496	49499	4	4	1.11E-03	7.00E-03	DMTN (+27)
HyperHier630	chr20	19866974	19867145	0.94	4.70	35100	100012	100016	5	5	4.84E-04	2.00E-03	RIN2 (-105)
HyperHier631	chr19	35940425	35940483	0.94	2.82	29032	96990	96992	3	3	3.03E-03	1.70E-02	FFAR2 (+1251), KRTDAP (+40979)
HyperHier632	chr1	223317364	223317548	0.94	3.76	5285	9041	9044	4	5	1.12E-03	7.00E-03	TLR5 (-6827), SUSD4 (+219945)
HyperHier633	chr5	176304988	176305077	0.94	2.82	46431	33761	33763	3	3	3.04E-03	1.70E-02	HK3 (+21300), UNC5A (+67555)
HyperHier634	chr10	72254314	72254335	0.94	2.82	7042	57584	57586	3	3	3.04E-03	1.70E-02	PALD1 (+15748), PRF1 (+108206)
HyperHier635	chr16	67516162	67516546	0.94	2.81	22640	85473	85475	3	3	3.06E-03	1.70E-02	ATP6V0D1 (-1214)
HyperHier636	chr17	33787402	33787739	0.94	2.81	24652	89020	89022	3	4	3.07E-03	1.70E-02	SLFN13 (-11715), SLFN12L (+27305)
HyperHier637	chr21	44863611	44864600	0.94	4.68	36495	102686	102690	5	5	4.94E-04	3.00E-03	SIK1 (-17098), HSF2BP (+215268)

HyperHier638	chr20	62327968	62328427	0.93	3.74	36106	101951	101954	4	4	1.15E-03	8.00E-03	TNFRSF6B (+177)
HyperHier639	chr14	104621467	104622128	0.93	4.67	18859	79057	79061	5	5	4.98E-04	4.00E-03	C14orf144 (-88743), KIF26A (+16738)
HyperHier640	chr12	6485537	6485916	0.93	2.80	12824	68113	68115	3	9	3.10E-03	1.80E-02	SCNN1A (-1337)
HyperHier641	chr6	89927233	89927651	0.93	2.80	48892	39480	39482	3	3	3.12E-03	1.80E-02	GABRR1 (-290)
HyperHier642	chr5	149887461	149887787	0.93	3.73	45847	32759	32762	4	4	1.16E-03	8.00E-03	NDST1 (-50)
HyperHier643	chr10	95326033	95326248	0.93	3.72	7512	58332	58335	4	4	1.16E-03	8.00E-03	FFAR4 (-281)
HyperHier644	chr10	11246612	11246763	0.93	2.79	6186	56086	56088	3	3	3.13E-03	1.80E-02	CELF2 (+186795), USP6NL (+327586)
HyperHier645	chr10	72362292	72362809	0.93	6.51	7047	57591	57597	7	7	1.44E-04	2.00E-03	PRF1 (-20)
HyperHier646	chr17	1633714	1634018	0.93	2.79	23708	87299	87301	3	3	3.14E-03	1.80E-02	SERPINF2 (-12264), WDR81 (+5612)
HyperHier647	chr4	2933715	2934188	0.93	3.72	41017	23713	23716	4	5	1.16E-03	8.00E-03	MFSD10 (+2233), ADD1 (+88368)
HyperHier648	chr6	170535724	170536124	0.93	3.71	50275	42054	42057	4	4	1.17E-03	8.00E-03	DLL1 (+63637), C6orf70 (+384203)
HyperHier649	chr11	4415193	4415245	0.93	2.79	9241	61935	61937	3	3	3.15E-03	1.80E-02	TRIM21 (-293)
HyperHier650	chr22	44568387	44568812	0.93	4.64	37383	104180	104184	5	5	5.10E-04	4.00E-03	PARVG (-236)
HyperHier651	chr1	156215418	156215951	0.93	3.71	3980	6849	6852	4	4	1.17E-03	8.00E-03	PAQR6 (+2158), BGLAP (+3932)
HyperHier652	chr15	45028083	45028270	0.93	3.71	19606	80277	80280	4	5	1.17E-03	8.00E-03	SORD (-287125), TRIM69 (+6991)
HyperHier653	chr11	14927004	14927549	0.93	3.71	9529	62410	62413	4	4	1.17E-03	8.00E-03	CYP2R1 (-13479), CALCA (+66623)
HyperHier654	chr13	20762982	20763457	0.93	2.78	15671	73259	73261	3	3	3.16E-03	1.80E-02	GJB2 (+699)
HyperHier655	chr16	1537775	1538803	0.93	5.56	21233	83155	83160	6	6	2.60E-04	3.00E-03	PTX4 (+179)
HyperHier656	chr2	102866879	102867466	0.93	3.70	32293	13240	13243	4	4	1.18E-03	8.00E-03	IL1RL1 (-60789), IL1RL2 (+63740)
HyperHier657	chr16	832828	833250	0.93	7.41	21064	82832	82839	8	9	8.84E-05	2.00E-03	MSLNL (-113)
HyperHier658	chr4	159131324	159131516	0.92	3.69	42892	27125	27128	4	4	1.20E-03	8.00E-03	TMEM144 (+19)
HyperHier659	chr1	41156730	41156980	0.92	2.77	2203	3677	3679	3	3	3.20E-03	1.80E-02	NFYC (-533)
HyperHier660	chr3	189349003	189349323	0.92	4.62	40596	22926	22930	5	5	5.21E-04	4.00E-03	TP63 (-53)
HyperHier661	chr17	37123767	37123949	0.92	4.61	24791	89276	89280	5	9	5.23E-04	4.00E-03	LASP1 (+97746), PLXDC1 (+184044)
HyperHier662	chr19	51220286	51220537	0.92	2.77	29889	98586	98588	3	3	3.22E-03	1.80E-02	SHANK1 (-217)
HyperHier663	chr11	69482043	69482597	0.92	3.69	10951	65062	65065	4	5	1.21E-03	8.00E-03	ORAOV1 (+7795), CCND1 (+26465)
HyperHier664	chr7	1428806	1428900	0.92	2.77	50491	42507	42509	3	3	3.22E-03	1.80E-02	MICALL2 (+70285), UNCX (+156310)
HyperHier665	chr3	107810361	107810791	0.92	5.53	39251	20585	20590	6	6	2.64E-04	3.00E-03	CD47 (-715)
HyperHier666	chr11	2193187	2193293	0.92	2.76	9138	61662	61664	3	4	3.23E-03	1.80E-02	TH (-205)
HyperHier667	chr1	9224003	9224376	0.92	2.76	760	1314	1316	3	3	3.24E-03	1.80E-02	H6PD (-70644), GPR157 (-34961)
HyperHier668	chr2	157291262	157292127	0.92	5.52	33123	14672	14677	6	6	2.65E-04	3.00E-03	GPD2 (-258)
HyperHier669	chr19	8372814	8373608	0.92	6.44	28211	95466	95472	7	7	1.52E-04	2.00E-03	CD320 (+28)
HyperHier670	chr11	67052509	67052992	0.92	3.67	10815	64777	64780	4	4	1.22E-03	8.00E-03	ANKRD13D (-4011)
HyperHier671	chr1	156046344	156046832	0.92	2.75	3960	6816	6818	3	3	3.27E-03	1.90E-02	MEX3A (+5201), RAB25 (+15637)
HyperHier672	chr3	10326540	10326774	0.92	2.75	37790	18087	18089	3	3	3.27E-03	2.00E-02	GHRL (+7974), ENSG00000272410 (+35601)
HyperHier673	chr10	131669406	131669630	0.92	2.75	8512	60305	60307	3	3	3.28E-03	2.00E-02	EBF3 (+92587), MGMT (+404070)
HyperHier674	chr4	3432342	3432546	0.92	2.75	41046	23771	23773	3	3	3.28E-03	2.00E-02	HGFAC (-11170), RGS12 (+116570)
HyperHier675	chr21	45232232	45232595	0.92	3.67	36510	102713	102716	4	4	1.23E-03	9.00E-03	AGPAT3 (-112475), RRP1 (+23020)
HyperHier676	chr8	144371537	144371779	0.92	2.75	56224	52980	52982	3	3	3.29E-03	2.00E-02	ZNF696 (-1901)
HyperHier677	chr2	65086822	65087288	0.91	2.74	31629	12144	12146	3	3	3.30E-03	2.00E-02	SERTAD2 (-206008), SLC1A4 (-129480)
HyperHier678	chr8	21905599	21905960	0.91	2.74	54215	49482	49484	3	4	3.30E-03	2.00E-02	DMTN (-10937), FGF17 (+5843)
HyperHier679	chr3	196065289	196065569	0.91	8.23	40756	23211	23219	9	9	5.79E-05	2.00E-03	TM4SF19 (-170)
HyperHier680	chr16	14403004	14403425	0.91	4.56	21704	83952	83956	5	5	5.46E-04	4.00E-03	MKL2 (+230070), PARN (+320909)
HyperHier681	chr11	1506936	1507347	0.91	3.65	9030	61389	61392	4	4	1.26E-03	9.00E-03	MOB2 (+834)
HyperHier682	chr17	3907349	3907994	0.91	3.64	23821	87500	87503	4	4	1.26E-03	9.00E-03	ATP2A3 (-40087), ZZEF1 (+138642)
HyperHier683	chr3	194408516	194409431	0.91	5.46	40708	23119	23124	6	9	2.75E-04	3.00E-03	LSG1 (-15768), XXYLT1 (+582922)
HyperHier684	chr8	22456092	22456421	0.91	3.64	54266	49566	49569	4	5	1.26E-03	9.00E-03	C8orf58 (-903)
HyperHier685	chr16	83171068	83171299	0.91	2.73	22947	85983	85985	3	3	3.35E-03	2.00E-02	HSBP1 (-670264), CDH13 (+510614)
HyperHier686	chr20	45947025	45947226	0.91	2.73	35657	101146	101148	3	3	3.35E-03	2.00E-02	ZMYND8 (+38288), EYA2 (+423863)
HyperHier687	chr6	161860203	161860359	0.91	2.73	49943	41480	41482	3	3	3.36E-03	2.10E-02	AGPAT4 (-165188)
HyperHier688	chr7	73720808	73721031	0.91	2.73	51884	45255	45257	3	3	3.37E-03	2.10E-02	GTF2IRD1 (-147380), CLIP2 (+17115)
HyperHier689	chr19	36004632	36004716	0.91	2.73	29036	97000	97002	3	4	3.37E-03	2.10E-02	DMKN (-120)
HyperHier690	chr3	64008007	64008387	0.91	4.54	38855	20020	20024	5	5	5.51E-04	4.00E-03	PSMD6 (+1039), ATXN7 (+157964)
HyperHier691	chr14	105619511	105619701	0.91	2.73	18965	79245	79247	3	3	3.37E-03	2.10E-02	GPR132 (-87839), JAG2 (+15555)
HyperHier692	chr16	764420	764794	0.91	2.72	21051	82809	82811	3	3	3.37E-03	2.10E-02	METRN (-508)
HyperHier693	chr10	134045514	134045913	0.91	3.63	8649	60566	60569	4	4	1.28E-03	1.00E-02	DPYSL4 (+45310), STK32C (+75730)
HyperHier694	chr19	5074616	5074758	0.91	2.72	28038	95176	95178	3	4	3.40E-03	2.10E-02	KDM4B (+105555), PTPRS (+266127)
HyperHier695	chr8	145018816	145018928	0.91	2.72	56306	53157	53159	3	3	3.41E-03	2.10E-02	EPPK1 (-66240), PLEC (+6172)

HyperHier696	chr2	234359654	234359783	0.90	2.71	34389	17072	17074	3	3	3.41E-03	2.10E-02	DGKD (+96566), USP40 (+114517)
HyperHier697	chr17	79228937	79229385	0.90	2.71	26546	92377	92379	3	3	3.42E-03	2.10E-02	C17orf89 (+16122), SLC38A10 (+39944)
HyperHier698	chr19	55586493	55587039	0.90	2.71	30146	99106	99108	3	4	3.42E-03	2.10E-02	EPS8L1 (-503)
HyperHier699	chr10	73533891	73534338	0.90	3.62	7083	57649	57652	4	4	1.30E-03	1.00E-02	C10orf54 (-860)
HyperHier700	chr13	113356724	113357112	0.90	2.71	16987	75722	75724	3	3	3.43E-03	2.10E-02	MCF2L (-266617), ATP11A (+12275)
HyperHier701	chr16	85465589	85465657	0.90	2.71	23078	86181	86183	3	3	3.44E-03	2.10E-02	GSE1 (-181199), KIAA0513 (+368805)
HyperHier702	chr14	24779793	24780926	0.90	11.74	17307	76271	76283	13	13	1.06E-05	1.00E-03	LTB4R (-2157), CIDEB (+241)
HyperHier703	chr12	49688844	49689212	0.90	2.71	13473	69190	69192	3	5	3.44E-03	2.10E-02	TROAP (-28034), PRPH (+1543)
HyperHier704	chr1	246728663	246728727	0.90	2.71	5837	9960	9962	3	3	3.44E-03	2.10E-02	CNST (-1053), TFB2M (+931)
HyperHier705	chr17	7341436	7341936	0.90	4.51	23994	87863	87867	5	8	5.65E-04	4.00E-03	FGF11 (-660)
HyperHier706	chr17	79374327	79374741	0.90	3.61	26575	92434	92437	4	4	1.31E-03	1.00E-02	ENSG00000171282 (+994)
HyperHier707	chr1	11795746	11796079	0.90	8.11	894	1531	1539	9	9	6.15E-05	2.00E-03	AGTRAP (-299)
HyperHier708	chr2	127413363	127413369	0.90	2.70	32700	13991	13993	3	3	3.48E-03	2.10E-02	GYPC (-143)
HyperHier709	chr21	34185927	34186122	0.90	2.69	36269	102227	102229	3	3	3.51E-03	2.20E-02	OLIG2 (-212218), C21orf62 (-5997)
HyperHier710	chr17	76975944	76976357	0.90	5.38	26331	91972	91977	6	6	2.90E-04	3.00E-03	LGALS3BP (+40)
HyperHier711	chr10	134362126	134362170	0.90	2.69	8681	60632	60634	3	3	3.51E-03	2.20E-02	INPP5A (+10824), NKX6-2 (+237408)
HyperHier712	chr17	77810509	77810912	0.90	2.69	26415	92128	92130	3	3	3.51E-03	2.20E-02	CBX8 (-39796), CBX4 (+2517)
HyperHier713	chr8	48648813	48649457	0.90	4.48	54790	50506	50510	5	5	5.76E-04	4.00E-03	CEBPD (+2513), SPIDR (+475968)
HyperHier714	chr13	50070550	50070766	0.90	2.69	16212	74245	74247	3	3	3.52E-03	2.20E-02	PHF11 (+561)
HyperHier715	chr11	130271767	130271903	0.90	2.69	12399	67414	67416	3	3	3.52E-03	2.20E-02	ZBTB44 (-87254), ADAMTS8 (+27053)
HyperHier716	chr14	104741786	104742172	0.90	2.69	18873	79088	79090	3	3	3.53E-03	2.20E-02	C14orf180 (-304042), C14orf144 (+31438)
HyperHier717	chr1	28843736	28844028	0.89	2.68	1723	2853	2855	3	3	3.54E-03	2.30E-02	RCC1 (-887)
HyperHier718	chr11	2241249	2241568	0.89	2.68	9146	61681	61683	3	3	3.54E-03	2.30E-02	TH (-48374), ASCL2 (+50773)
HyperHier719	chr12	133022677	133022955	0.89	3.58	15577	73086	73089	4	6	1.35E-03	1.00E-02	GALNT9 (-332243), MUC8 (+27910)
HyperHier720	chr10	3823758	3824687	0.89	6.25	6056	55841	55847	7	7	1.70E-04	2.00E-03	PITRM1 (-609220), KLF6 (+3244)
HyperHier721	chr20	57267176	57267680	0.89	3.57	35828	101448	101451	4	4	1.36E-03	1.00E-02	NPEPL1 (-241)
HyperHier722	chr17	80988390	80988709	0.89	2.68	26782	92851	92853	3	3	3.56E-03	2.40E-02	ZNF750 (-190096), B3GNTL1 (+21136)
HyperHier723	chr2	231090329	231090745	0.89	4.46	34268	16873	16877	5	5	5.89E-04	4.00E-03	SP140 (+91)
HyperHier724	chr19	39360313	39361447	0.89	4.45	29183	97301	97305	5	5	5.92E-04	4.00E-03	HNRNPL (-19907), RINL (+8039)
HyperHier725	chr9	137772544	137772580	0.89	2.67	57731	55136	55138	3	4	3.61E-03	2.50E-02	FCN2 (-96)
HyperHier726	chr6	30686922	30687221	0.89	3.55	47644	36599	36602	4	4	1.39E-03	1.10E-02	MDC1 (-1406), TUBB (-906)
HyperHier727	chr22	32026779	32026975	0.89	4.44	37037	103586	103590	5	5	5.99E-04	4.00E-03	PISD (-2670)
HyperHier728	chr2	219233446	219233650	0.89	2.66	33987	16303	16305	3	3	3.62E-03	2.50E-02	TMBIM1 (-81976), SLC11A1 (-13204)
HyperHier729	chr8	22735111	22735478	0.89	4.44	54283	49599	49603	5	5	5.99E-04	4.00E-03	EGR3 (-184480), PEBP4 (+50126)
HyperHier730	chr6	11111894	11112117	0.89	2.66	46997	34839	34841	3	3	3.62E-03	2.50E-02	ERVFRD-1 (-41)
HyperHier731	chr9	139871955	139872395	0.89	3.55	57912	55387	55390	4	6	1.40E-03	1.10E-02	PTGDS (+218)
HyperHier732	chr5	149867577	149868855	0.89	3.54	45844	32753	32756	4	4	1.40E-03	1.10E-02	RPS14 (-38906), NDST1 (-19458)
HyperHier733	chr6	10555808	10556326	0.89	3.54	46983	34784	34787	4	5	1.40E-03	1.10E-02	GCNT6 (-77926), GCNT2 (+27478)
HyperHier734	chr11	1948933	1949113	0.89	4.43	9095	61514	61518	5	5	6.05E-04	4.00E-03	MRPL23 (-19485), TNNT3 (+8231)
HyperHier735	chr3	4794020	4794082	0.88	2.65	37706	17930	17932	3	3	3.68E-03	2.90E-02	BHLHE40 (-226750), ITPR1 (+259019)
HyperHier736	chr10	131843597	131844047	0.88	3.52	8529	60361	60364	4	5	1.43E-03	1.30E-02	GLRX3 (-90841), EBF3 (-81717)
HyperHier737	chr1	155043883	155044345	0.88	5.29	3909	6729	6734	6	6	3.05E-04	3.00E-03	EFNA3 (-7234), EFNA4 (+7877)
HyperHier738	chr1	247681242	247681439	0.88	2.64	5861	10012	10014	3	6	3.71E-03	3.10E-02	OR2B11 (-66033), OR2C3 (+15800)
HyperHier739	chr6	170559721	170559869	0.88	2.64	50280	42062	42064	3	3	3.71E-03	3.10E-02	DLL1 (+39766), C6orf70 (+408074)
HyperHier740	chr18	61604237	61604367	0.88	2.64	27317	93927	93929	3	3	3.72E-03	3.10E-02	HMSD (-12233), SERPINB10 (+29077)
HyperHier741	chr6	30029929	30030069	0.88	3.52	47563	36242	36245	4	4	1.44E-03	1.40E-02	PPP1R11 (-4866), ZNRD1 (+968)
HyperHier742	chr10	6185502	6186015	0.88	2.64	6107	55931	55933	3	3	3.73E-03	3.20E-02	PFKFB3 (-59135), RBM17 (+54450)
HyperHier743	chr17	78829626	78829930	0.88	2.63	26494	92272	92274	3	3	3.73E-03	3.20E-02	CHMP6 (-135863), RPTOR (+310710)
HyperHier744	chr6	42883504	42883762	0.88	3.51	48364	38514	38517	4	4	1.45E-03	1.40E-02	PTCRA (-94)
HyperHier745	chr16	89469541	89469845	0.88	2.63	23494	86925	86927	3	3	3.74E-03	3.20E-02	ANKRD11 (+87276), ZNF778 (+185575)
HyperHier746	chr16	88844415	88845204	0.88	5.27	23407	86755	86760	6	6	3.11E-04	3.00E-03	PIEZO1 (+6809), CTU2 (+71939)
HyperHier747	chr19	43856594	43857074	0.88	2.63	29391	97650	97652	3	3	3.76E-03	3.20E-02	PSG9 (-83154), TEX101 (-48814)
HyperHier748	chr20	60456296	60456503	0.88	2.63	35887	101581	101583	3	3	3.76E-03	3.40E-02	TAF4 (+184466), CDH4 (+628918)
HyperHier749	chr13	49107693	49108353	0.87	2.62	16194	74220	74222	3	3	3.78E-03	3.50E-02	RCBTB2 (-654)
HyperHier750	chr6	25027586	25027714	0.87	2.62	47221	35186	35188	3	5	3.78E-03	3.50E-02	LRRC16A (-252006), FAM65B (-116455)
HyperHier751	chr1	26527870	26527928	0.87	2.62	1609	2636	2638	3	3	3.80E-03	3.50E-02	CEP85 (-32794), CATSPER4 (+10847)
HyperHier752	chr1	197745025	197745238	0.87	2.62	4652	7979	7981	3	3	3.80E-03	3.50E-02	DENND1B (-677)
HyperHier753	chr8	143407432	143408047	0.87	2.62	56136	52814	52816	3	3	3.80E-03	3.50E-02	TSNARE1 (+76803)

HyperHier754	chr3	193987426	193987757	0.87	3.49	40684	23074	23077	4	4	1.48E-03	1.50E-02	CPN2 (+84465), HES1 (+133658)
HyperHier755	chr20	52199520	52199778	0.87	4.36	35736	101266	101270	5	5	6.42E-04	6.00E-03	ZNF217 (+10729), TSHZ2 (+610703)
HyperHier756	chr12	54778312	54779175	0.87	3.49	13797	69875	69878	4	4	1.48E-03	1.50E-02	GPR84 (-20473), ZNF385A (+6338)
HyperHier757	chr10	129797760	129797840	0.87	2.61	8463	60212	60214	3	3	3.82E-03	3.60E-02	PTPRE (+92475), MKI67 (+126849)
HyperHier758	chr17	79109729	79109910	0.87	3.48	26534	92349	92352	4	5	1.49E-03	1.50E-02	AATK (+29997), BAIAP2 (+100858)
HyperHier759	chr1	1897585	1897785	0.87	2.61	168	343	345	3	3	3.83E-03	3.60E-02	GABRD (-53095), TMEM52 (-46973)
HyperHier760	chr13	114810631	114810794	0.87	2.61	17099	75923	75925	3	3	3.84E-03	3.60E-02	GAS6 (-243673), RASA3 (+87373)
HyperHier761	chr9	139237366	139237503	0.87	2.61	57841	55289	55291	3	3	3.84E-03	3.60E-02	GPSM1 (+15503), DNLZ (+20806)
HyperHier762	chr19	55219519	55219964	0.87	2.61	30129	99077	99079	3	3	3.84E-03	3.60E-02	KIR3DL3 (-16242), LILRB4 (+46163)
HyperHier763	chr16	89168508	89168963	0.87	2.61	23461	86877	86879	3	4	3.84E-03	3.60E-02	CDH15 (-69439), ACSF3 (+8519)
HyperHier764	chr8	145024929	145025760	0.87	7.83	56308	53161	53169	9	9	7.02E-05	2.00E-03	PLEC (-301)
HyperHier765	chr20	61508481	61508590	0.87	2.61	35969	101723	101725	3	3	3.85E-03	3.60E-02	TCFL5 (-15421), DIDO1 (+60738)
HyperHier766	chr7	4778839	4779342	0.87	5.22	50710	42921	42926	6	6	3.23E-04	4.00E-03	AP5Z1 (-36162), FOXK1 (+57151)
HyperHier767	chr20	896876	897050	0.87	2.61	34873	99588	99590	3	3	3.85E-03	3.60E-02	ANGPT4 (-3)
HyperHier768	chr1	1058077	1058658	0.87	2.61	58	113	115	3	3	3.86E-03	3.60E-02	TTLL10 (-50915), C1orf159 (-6890)
HyperHier769	chr12	56040025	56040336	0.87	4.34	13826	69921	69925	5	5	6.53E-04	6.00E-03	METTL7B (-35149), OR10P1 (+9537)
HyperHier770	chr3	183543680	183543862	0.87	2.60	40463	22707	22709	3	3	3.86E-03	3.60E-02	MAP6D1 (-405)
HyperHier771	chr2	85637673	85637851	0.87	3.47	32042	12839	12842	4	4	1.51E-03	1.50E-02	CAPG (-86)
HyperHier772	chr1	54619445	54619895	0.87	2.60	2604	4391	4393	3	3	3.87E-03	3.60E-02	CDCP2 (-227)
HyperHier773	chr11	57232331	57232556	0.87	3.47	10299	63900	63903	4	4	1.52E-03	1.50E-02	RTN4RL2 (+4422), SLC43A1 (+50815)
HyperHier774	chr11	2846681	2847019	0.86	3.46	9187	61779	61782	4	4	1.53E-03	1.70E-02	CDKN1C (+60261), KCNQ1 (+380629)
HyperHier775	chr11	67764654	67765204	0.86	4.32	10864	64879	64883	5	5	6.65E-04	6.00E-03	ALDH3B2 (-322827), UNC93B1 (+6664)
HyperHier776	chr2	242813634	242814898	0.86	4.32	34822	17822	17826	5	5	6.66E-04	6.00E-03	CXXC11 (+2385)
HyperHier777	chr5	145236139	145236262	0.86	2.59	45728	32535	32537	3	3	3.96E-03	4.20E-02	PRELID2 (-21334), GRXCR2 (+16330)
HyperHier778	chr1	9884565	9885517	0.86	8.62	808	1387	1396	10	10	4.75E-05	2.00E-03	CLSTN1 (-457)
HyperHier779	chr1	243645911	243646787	0.86	6.03	5780	9878	9884	7	7	1.93E-04	3.00E-03	SDCCAG8 (+226991), AKT3 (+367081)
HyperHier780	chr16	69966815	69967212	0.86	5.16	22718	85618	85623	6	6	3.35E-04	4.00E-03	CLEC18A (-18069), WWP2 (+170805)
HyperHier781	chr20	30073209	30073576	0.86	5.16	35241	100359	100364	6	6	3.35E-04	4.00E-03	HM13 (-28838), REM1 (+10297)
HyperHier782	chr6	30124214	30124299	0.86	3.44	47587	36383	36386	4	4	1.57E-03	1.90E-02	TRIM10 (+4454), TRIM40 (+19470)
HyperHier783	chr2	240259002	240259213	0.86	2.57	34662	17536	17538	3	3	4.01E-03	4.30E-02	HDAC4 (+63535), TWIST2 (+502435)
HyperHier784	chr22	46508454	46508604	0.86	4.29	37474	104359	104363	5	5	6.86E-04	8.00E-03	WNT7B (-135520), PPARA (-37970)
HyperHier785	chr16	891283	891605	0.86	3.43	21081	82873	82876	4	4	1.58E-03	2.00E-02	GNG13 (-40711), LMF1 (+129555)
HyperHier786	chr14	104668772	104668921	0.86	3.43	18865	79069	79072	4	4	1.58E-03	2.00E-02	C14orf144 (-41694), KIF26A (+63787)
HyperHier787	chr12	122444450	122444621	0.86	2.57	15083	72196	72198	3	3	4.02E-03	4.30E-02	BCL7A (-12792), WDR66 (+88768)
HyperHier788	chr1	248902767	248903325	0.86	4.28	5873	10049	10053	5	5	6.91E-04	8.00E-03	ZNF672 (-229433), OR14I1 (-57417)
HyperHier789	chr17	81039747	81039990	0.86	2.57	26790	92866	92868	3	5	4.04E-03	4.40E-02	METRNL (+2302)
HyperHier790	chr2	10470961	10471388	0.85	2.56	30681	10596	10598	3	3	4.06E-03	4.40E-02	HPCAL1 (+27349), ODC1 (+117455)
HyperHier791	chr3	13060741	13060974	0.85	3.42	37885	18256	18259	4	4	1.59E-03	2.00E-02	IQSEC1 (-51690), NUP210 (+400951)
HyperHier792	chr19	836606	836716	0.85	2.56	27591	94420	94422	3	3	4.07E-03	4.40E-02	PRTN3 (-4302)
HyperHier793	chr16	85607562	85608058	0.85	2.56	23100	86223	86225	3	3	4.08E-03	4.40E-02	GSE1 (-39012), KIAA0513 (+510992)
HyperHier794	chr11	61159687	61159739	0.85	2.56	10413	64114	64116	3	5	4.09E-03	4.40E-02	TMEM216 (+554)
HyperHier795	chr17	6358363	6358599	0.85	3.41	23922	87677	87680	4	5	1.61E-03	2.00E-02	FAM64A (+10720), ACKR6 (+101333)
HyperHier796	chr7	27141388	27142100	0.85	2.55	51035	43632	43634	3	28	4.11E-03	4.40E-02	HOXA2 (+686)
HyperHier797	chr11	67273431	67273507	0.85	2.55	10844	64836	64838	3	3	4.11E-03	4.40E-02	PITPNM1 (-626)
HyperHier798	chr8	142297121	142297256	0.85	2.55	56058	52696	52698	3	3	4.14E-03	4.40E-02	SLC45A4 (-58516), GPR20 (+80178)
HyperHier799	chr12	6756377	6756678	0.85	3.39	12851	68164	68167	4	7	1.64E-03	2.10E-02	ACRBP (+98)
HyperHier800	chr2	238578376	238578491	0.85	2.54	34574	17374	17376	3	3	4.17E-03	4.70E-02	RAB17 (-78698), LRRFIP1 (-22548)
HyperHier801	chr6	28829931	28830041	0.85	3.38	47454	35840	35843	4	4	1.65E-03	2.10E-02	SCAND3 (-274874), TRIM27 (+61780)
HyperHier802	chr5	96078430	96078834	0.85	2.54	44819	30730	30732	3	3	4.19E-03	4.80E-02	ERAP1 (+65171), CAST (+80691)
HyperHier803	chr4	24981695	24981807	0.85	2.54	41584	24760	24762	3	5	4.19E-03	4.80E-02	LGI2 (+50750), SOD3 (+184666)
HyperHier804	chr3	46448084	46449313	0.85	7.61	38378	19110	19118	9	9	7.90E-05	2.00E-03	ACKR5 (-350)
HyperHier805	chr8	144255650	144255688	0.84	2.53	56207	52951	52953	3	3	4.20E-03	4.80E-02	GPIHBP1 (-39399), LY6H (-13541)
HyperHier806	chr17	48473757	48474021	0.84	2.53	25509	90659	90661	3	3	4.21E-03	4.80E-02	LRRC59 (+1025), EME1 (+23308)
HyperHier807	chr5	141062313	141062571	0.84	2.53	45629	32407	32409	3	3	4.22E-03	4.90E-02	ARAP3 (-654)
HyperHier808	chr2	242754333	242754514	0.84	2.53	34806	17789	17791	3	3	4.23E-03	4.90E-02	NEU4 (+2329), PDCD1 (+46636)
HyperHier809	chr1	1173264	1173622	0.84	2.53	83	163	165	3	4	4.24E-03	4.90E-02	B3GALT6 (+5814), FAM132A (+8659)
HyperHier810	chr18	74845154	74845829	0.84	3.37	27390	94078	94081	4	4	1.67E-03	2.10E-02	MBP (-767)
HyperHier811	chr4	183795763	183795822	0.84	2.53	43112	27547	27549	3	3	4.25E-03	4.90E-02	DCTD (+42739), TENM3 (+631211)

HyperHier812	chr22	25595589	25595840	0.84	4.20	36908	103373	103377	5	5	7.42E-04	9.00E-03	CRYBB3 (-102)
HyperHier813	chr19	4539943	4540333	0.84	2.52	27998	95106	95108	3	3	4.26E-03	5.00E-02	PLIN5 (-4902), LRG1 (+348)
HyperHier814	chr12	7070562	7070732	0.84	2.52	12887	68225	68227	3	3	4.28E-03	5.00E-02	PHB2 (+9341), PTPN6 (+10213)
HyperHier815	chr12	109203581	109203758	0.84	2.52	14642	71270	71272	3	3	4.28E-03	5.00E-02	CORO1C (-78298), SSH1 (+47696)
HyperHier816	chr11	66103386	66104485	0.84	9.23	10742	64658	64668	11	11	3.19E-05	2.00E-03	RIN1 (+64)
HyperHier817	chr17	79429330	79429729	0.84	2.51	26598	92485	92487	3	3	4.30E-03	5.10E-02	ACTG1 (+50277), ENSG00000171282 (+55990)
HyperHier818	chr19	852114	852311	0.84	2.51	27599	94442	94444	3	4	4.31E-03	5.10E-02	CFD (-7240), ELANE (+1199)
HyperHier819	chr17	3824384	3824497	0.84	2.51	23814	87490	87492	3	3	4.31E-03	5.10E-02	P2RX1 (-4647)
HyperHier820	chr18	77545028	77545223	0.84	2.51	27471	94225	94227	3	3	4.33E-03	5.10E-02	KCNG2 (-78542), CTDP1 (+105325)
HyperHier821	chr14	104171259	104172109	0.84	5.02	18827	78998	79003	6	6	3.76E-04	5.00E-03	XRCC3 (+6757), KLC1 (+76106)
HyperHier822	chr20	748992	749620	0.84	5.85	34865	99558	99564	7	7	2.14E-04	3.00E-03	SLC52A3 (-175)
HyperHier823	chr17	78851149	78851262	0.84	2.51	26496	92277	92279	3	4	4.35E-03	5.20E-02	CHMP6 (-114435), RPTOR (+332138)
HyperHier824	chr1	153513810	153514482	0.84	3.34	3825	6570	6573	4	4	1.73E-03	2.20E-02	S100A5 (+95)
HyperHier825	chr1	3394709	3395258	0.83	3.34	427	805	808	4	4	1.74E-03	2.20E-02	ARHGEF16 (+23994), MEGF6 (+133075)
HyperHier826	chr16	89151047	89151343	0.83	2.50	23455	86859	86861	3	3	4.37E-03	5.30E-02	CBFA2T3 (-107583), ACSF3 (-9022)
HyperHier827	chr10	5882304	5882576	0.83	2.50	6095	55909	55911	3	3	4.37E-03	5.30E-02	FBXO18 (-53909), GDI2 (-26928)
HyperHier828	chr6	30070074	30070738	0.83	7.50	47571	36310	36318	9	9	8.35E-05	2.00E-03	RNF39 (-26742), TRIM31 (+10477)
HyperHier829	chr7	157695747	157695878	0.83	2.50	53546	48326	48328	3	3	4.39E-03	5.40E-02	DNAJB6 (+566153), PTPRN2 (+684558)
HyperHier830	chr7	2773010	2773127	0.83	2.50	50649	42812	42814	3	4	4.40E-03	5.40E-02	AMZ1 (+53913), GNA12 (+110889)
HyperHier831	chr5	134735544	134735914	0.83	4.99	45265	31479	31484	6	6	3.80E-04	5.00E-03	H2AFY (-417)
HyperHier832	chr2	27719501	27720122	0.83	3.32	31077	11256	11259	4	4	1.76E-03	2.50E-02	FNDC4 (-1700), GCKR (+103)
HyperHier833	chr11	63448356	63448455	0.83	2.49	10536	64311	64313	3	3	4.42E-03	5.40E-02	RTN3 (-549)
HyperHier834	chr5	126564606	126564742	0.83	2.49	45126	31220	31222	3	3	4.42E-03	5.40E-02	C5orf63 (-155490), MEGF10 (-61877)
HyperHier835	chr7	155640537	155640784	0.83	2.49	53374	47975	47977	3	3	4.43E-03	5.70E-02	SHH (-35694), C7orf13 (+792687)
HyperHier836	chr6	15501652	15502099	0.83	2.49	47089	34950	34952	3	3	4.45E-03	5.70E-02	DTNBP1 (+161397), JARID2 (+255349)
HyperHier837	chr13	113622539	113622750	0.83	4.97	17009	75762	75767	6	6	3.87E-04	6.00E-03	MCF2L (-890)
HyperHier838	chr12	4397947	4398508	0.83	2.48	12751	67976	67978	3	3	4.46E-03	5.80E-02	C12orf5 (-32143), CCND2 (+15290)
HyperHier839	chr3	129024601	129024712	0.83	2.48	39700	21286	21288	3	3	4.46E-03	5.80E-02	H1FX (+10463), COPG1 (+56208)
HyperHier840	chr7	150885084	150885458	0.83	2.48	53206	47627	47629	3	6	4.47E-03	5.80E-02	ASB10 (-1029)
HyperHier841	chr12	54942853	54943009	0.83	2.48	13810	69897	69899	3	3	4.47E-03	5.80E-02	PDE1B (-203)
HyperHier842	chr13	113689422	113689955	0.83	3.31	17028	75802	75805	4	5	1.79E-03	3.00E-02	F7 (-70432), MCF2L (+66154)
HyperHier843	chr17	80345221	80345367	0.83	2.48	26719	92734	92736	3	3	4.48E-03	5.90E-02	HEXDC (-30900), UTS2R (+13141)
HyperHier844	chr7	130125756	130125836	0.83	3.31	52822	46946	46949	4	14	1.79E-03	3.00E-02	CEP41 (-44718), MEST (-6138)
HyperHier845	chr11	70563131	70563792	0.83	3.30	11039	65244	65247	4	8	1.79E-03	3.00E-02	SHANK2 (+294910), CTTN (+318815)
HyperHier846	chr5	176543901	176544135	0.83	3.30	46443	33776	33779	4	5	1.80E-03	3.00E-02	NSD1 (-16908), FGFR4 (+30131)
HyperHier847	chr14	105125984	105126372	0.83	2.48	18914	79158	79160	3	3	4.50E-03	5.90E-02	TMEM179 (-54194), INF2 (-29796)
HyperHier848	chr3	50376000	50376216	0.83	3.30	38523	19434	19437	4	4	1.80E-03	3.00E-02	TUSC2 (-10434), RASSF1 (+2164)
HyperHier849	chr12	4488893	4489155	0.83	2.48	12755	67986	67988	3	6	4.51E-03	5.90E-02	FGF23 (-130)
HyperHier850	chr10	134664471	134664800	0.82	2.47	8712	60711	60713	3	3	4.54E-03	6.20E-02	GPR123 (-236773), NKX6-2 (-65080)
HyperHier851	chr17	80292079	80292252	0.82	2.47	26710	92706	92708	3	3	4.55E-03	6.20E-02	SECTM1 (-292)
HyperHier852	chr3	13678918	13679636	0.82	5.76	37913	18325	18331	7	7	2.28E-04	6.00E-03	FBLN2 (+88646), WNT7A (+242341)
HyperHier853	chr13	31019673	31020218	0.82	3.29	15927	73748	73751	4	5	1.82E-03	3.30E-02	KATNAL1 (-138325), HMGB1 (+171788)
HyperHier854	chr16	31548640	31549141	0.82	2.47	22102	84542	84544	3	3	4.55E-03	6.20E-02	ZNF720 (-175664), AHSP (+9706)
HyperHier855	chr8	144408126	144408900	0.82	3.29	56227	52989	52992	4	4	1.83E-03	3.30E-02	TOP1MT (+8553), ZNF696 (+34954)
HyperHier856	chr13	113296192	113296447	0.82	3.29	16981	75713	75716	4	4	1.83E-03	3.30E-02	TUBGCP3 (-53839), ATP11A (-48323)
HyperHier857	chr1	152595322	152596177	0.82	5.74	3772	6489	6495	7	7	2.31E-04	6.00E-03	LCE3A (-171)
HyperHier858	chr21	47456315	47456446	0.82	2.46	36656	102941	102943	3	3	4.60E-03	6.20E-02	COL6A2 (-61630), COL6A1 (+54730)
HyperHier859	chr17	19628012	19628421	0.82	3.28	24356	88510	88513	4	4	1.84E-03	3.40E-02	SLC47A2 (-8298), ALDH3A1 (+20555)
HyperHier860	chr1	16091100	16091599	0.82	2.45	1054	1771	1773	3	4	4.62E-03	6.30E-02	FBLIM1 (+356)
HyperHier861	chr10	6214016	6214079	0.82	2.45	6111	55937	55939	3	3	4.63E-03	6.30E-02	PFKFB3 (-30846), RBM17 (+82739)
HyperHier862	chr13	24269867	24270321	0.82	2.45	15747	73408	73410	3	3	4.63E-03	6.30E-02	TNFRSF19 (+116595), MIPEP (+193464)
HyperHier863	chr21	46077715	46077731	0.82	2.45	36561	102792	102794	3	5	4.64E-03	6.30E-02	KRTAP12-4 (-3147), KRTAP12-3 (-126)
HyperHier864	chr10	88717364	88717926	0.82	2.45	7402	58130	58132	3	6	4.65E-03	6.40E-02	SNCG (-768), MMRN2 (-273)
HyperHier865	chr9	126101872	126102244	0.81	2.44	57291	54574	54576	3	3	4.69E-03	6.70E-02	STRBP (-71203), CRB2 (-16481)
HyperHier866	chr12	133161835	133162043	0.81	2.44	15601	73123	73125	3	3	4.71E-03	6.80E-02	MUC8 (-111213), P2RX2 (-33464)
HyperHier867	chr2	55450737	55450874	0.81	2.44	31525	11964	11966	3	3	4.71E-03	6.80E-02	RTN4 (-173126), RPS27A (-8732)
HyperHier868	chr6	30168645	30169069	0.81	2.44	47594	36436	36438	3	3	4.72E-03	7.00E-02	TRIM26 (+12286), TRIM15 (+37864)
HyperHier869	chr5	191127	191486	0.81	3.25	43330	27925	27928	4	10	1.91E-03	3.80E-02	SDHA (-27049), PLEKHG4B (+50934)

HyperHier870	chr8	127568650	127569163	0.81	2.43	55823	52345	52347	3	7	4.76E-03	7.30E-02	FAM84B (+1731)
HyperHier871	chr1	2838805	2839247	0.81	4.05	319	610	614	5	5	8.49E-04	1.80E-02	MMEL1 (-274597), ACTRT2 (-99020)
HyperHier872	chr19	56004925	56005437	0.81	2.43	30199	99203	99205	3	3	4.77E-03	7.50E-02	SSC5D (+5247), SBK2 (+43275)
HyperHier873	chr3	3151795	3152038	0.81	2.43	37692	17908	17910	3	3	4.77E-03	7.50E-02	IL5RA (+141)
HyperHier874	chr19	10213271	10213722	0.81	2.43	28281	95597	95599	3	3	4.77E-03	7.50E-02	PPAN-P2RY11 (-3574), PPAN (-3547), ANGPTL6 (-25)
HyperHier875	chr1	230849801	230850299	0.81	2.43	5545	9493	9495	3	3	4.78E-03	7.50E-02	AGT (-7)
HyperHier876	chr1	1957073	1957805	0.81	4.04	180	370	374	5	5	8.54E-04	1.80E-02	PRKCZ (-24470), GABRD (+6659)
HyperHier877	chr16	89118603	89119709	0.81	6.46	23452	86849	86856	8	8	1.49E-04	8.00E-03	CBFA2T3 (-75544), ACSF3 (-41061)
HyperHier878	chr16	29195900	29196472	0.81	2.42	21973	84325	84327	3	3	4.81E-03	7.70E-02	LAT (+200039), NPIPB11 (+219164)
HyperHier879	chr16	3210066	3210357	0.81	2.42	21402	83469	83471	3	3	4.84E-03	8.00E-02	OR1F1 (-44035), CASP16 (+15968)
HyperHier880	chr14	105147537	105147781	0.80	2.41	18918	79164	79166	3	3	4.88E-03	8.50E-02	TMEM179 (-75675), INF2 (-8315)
HyperHier881	chr17	79377272	79378207	0.80	4.01	26578	92440	92444	5	5	8.83E-04	2.60E-02	ENSG00000171282 (+4200), ACTG1 (+102067)
HyperHier882	chr2	98852895	98852938	0.80	2.40	32208	13111	13113	3	3	4.91E-03	8.70E-02	TMEM131 (-240563), CNGA3 (-109701)
HyperHier883	chr15	89153667	89154196	0.80	3.20	20573	81883	81886	4	4	2.00E-03	4.70E-02	DET1 (-64048), AEN (-10595)
HyperHier884	chr7	1025760	1026310	0.80	3.20	50390	42258	42261	4	5	2.01E-03	4.70E-02	GPR146 (-68886), CYP2W1 (+3200)
HyperHier885	chr19	10405955	10406364	0.80	2.39	28299	95628	95630	3	3	4.97E-03	8.80E-02	ICAM5 (+5503), ZGLP1 (+14396)
HyperHier886	chr8	10530053	10530148	0.80	2.39	53985	49046	49048	3	3	4.97E-03	8.80E-02	C8orf74 (-46)
HyperHier887	chr16	1593178	1593557	0.80	3.19	21243	83187	83190	4	4	2.02E-03	4.70E-02	TMEM204 (+9794), IFT140 (+68743)
HyperHier888	chr5	179588440	179588673	0.80	2.39	46593	34047	34049	3	4	4.98E-03	8.80E-02	RASGEF1C (-23208), MAPK9 (+130542)
HyperHier889	chr16	4732911	4733181	0.80	2.39	21509	83657	83659	3	3	5.02E-03	9.10E-02	NUDT16L1 (-10649), MGRN1 (+58255)
HyperHier890	chr1	113265679	113265753	0.79	2.38	3427	5867	5869	3	3	5.02E-03	9.10E-02	FAM19A3 (+2675), SLC16A1 (+232969)
HyperHier891	chr17	17255427	17255771	0.79	2.38	24272	88389	88391	3	3	5.07E-03	9.40E-02	MED9 (-124701), NT5M (+48950)
HyperHier892	chr1	27894955	27895490	0.79	2.38	1686	2798	2800	3	3	5.08E-03	9.40E-02	WASF2 (-78554), AHDC1 (+34920)
HyperHier893	chr16	31487813	31488238	0.79	2.38	22094	84529	84531	3	3	5.08E-03	9.40E-02	SLC5A2 (-6413), TGFB1I1 (+4646)
HyperHier894	chr17	4835476	4835627	0.79	2.37	23875	87598	87600	3	3	5.09E-03	9.40E-02	GP1BA (-40)
HyperHier895	chr1	2995301	2995604	0.79	2.37	353	668	670	3	3	5.11E-03	9.40E-02	ARHGEF16 (-375537), PRDM16 (+9678)
HyperHier896	chr6	29599012	29599390	0.79	7.90	47518	36039	36048	10	11	6.88E-05	6.00E-03	GABBR1 (+1761), OR2H2 (+43518)
HyperHier897	chr2	1286394	1286641	0.79	3.16	30418	10190	10193	4	4	2.09E-03	4.90E-02	TPO (-130715), SNTG2 (+339964)
HyperHier898	chr8	884647	884842	0.79	2.37	53710	48598	48600	3	3	5.12E-03	9.50E-02	DLGAP2 (-564787), TDRP (-388964)
HyperHier899	chr10	135171713	135171746	0.79	2.37	8822	60952	60954	3	3	5.12E-03	9.50E-02	FUOM (-201)
HyperHier900	chr12	16499963	16500185	0.79	2.37	13090	68533	68535	3	4	5.15E-03	9.60E-02	MGST1 (-638)
HyperHier901	chr7	138720989	138721065	0.79	2.36	52957	47197	47199	3	3	5.16E-03	9.60E-02	KIAA1549 (-54963), ZC3HAV1 (+73367)
HyperHier902	chr7	157348161	157348266	0.79	2.36	53473	48189	48191	3	3	5.17E-03	9.60E-02	DNAJB6 (+218554)
HyperHier903	chr7	4746131	4746462	0.79	2.36	50703	42900	42902	3	3	5.19E-03	9.60E-02	AP5Z1 (-68956), FOXK1 (+24357)
HyperHier904	chr17	46676099	46676375	0.79	2.36	25360	90335	90337	3	4	5.19E-03	9.60E-02	HOXB5 (-4914)
HyperHier905	chr16	8738240	8738531	0.79	3.15	21595	83776	83779	4	4	2.13E-03	5.10E-02	ABAT (-68440), METTL22 (+22841)
HyperHier906	chr3	100712322	100712345	0.79	2.36	39206	20534	20536	3	3	5.20E-03	9.60E-02	ABI3BP (+25)
HyperHier907	chr17	80372493	80372912	0.79	3.14	26722	92741	92744	4	4	2.13E-03	5.10E-02	HEXDC (-3491)
HyperHier908	chr1	36785859	36788416	0.79	7.86	1998	3316	3325	10	10	6.95E-05	6.00E-03	EVA1B (+2617), THRAP3 (+97121)
HyperHier909	chr14	62035431	62035585	0.79	2.36	17804	77320	77322	3	3	5.21E-03	9.70E-02	HIF1A (-128832), PRKCH (+247073)
HyperHier910	chr9	141016791	141017457	0.79	2.36	58015	55541	55543	3	3	5.22E-03	9.80E-02	CACNA1B (+244883)
HyperHier911	chr17	1133546	1133706	0.79	2.36	23652	87192	87194	3	3	5.22E-03	9.80E-02	ABR (-50458), BHLHA9 (-40227)
HyperHier912	chr15	40802888	40803416	0.78	2.35	19496	80109	80111	3	3	5.24E-03	9.90E-02	RPUSD2 (-58347), CHST14 (+39992)
HyperHier913	chr1	180922636	180923569	0.78	3.92	4516	7765	7769	5	6	9.65E-04	3.00E-02	STX6 (+68944), XPR1 (+321963)
HyperHier914	chr6	31804078	31804883	0.78	3.13	47784	37107	37110	4	4	2.16E-03	5.30E-02	HSPA1B (+8969), NEU1 (+26202)
HyperHier915	chr16	1393584	1394089	0.78	3.13	21210	83106	83109	4	4	2.17E-03	5.30E-02	TSR3 (+8075), BAIAP3 (+9332)
HyperHier916	chr13	20797356	20797693	0.78	3.13	15676	73273	73276	4	4	2.17E-03	5.30E-02	GJB2 (-33606), GJB6 (+9009)
HyperHier917	chr2	25499619	25500416	0.78	3.91	30974	11082	11086	5	5	9.73E-04	3.00E-02	POMC (-108246), DNMT3A (+65441)
HyperHier918	chr10	81002480	81003657	0.78	3.91	7265	57903	57907	5	5	9.73E-04	3.00E-02	PPIF (-104165), ZMIZ1 (+174277)
HyperHier919	chr11	65314454	65315625	0.78	6.25	10683	64543	64550	8	11	1.70E-04	1.10E-02	LTBP3 (+10659), SCYL1 (+22492)
HyperHier920	chr11	62621178	62621791	0.78	3.90	10504	64259	64263	5	5	9.78E-04	3.00E-02	SLC3A2 (-2098)
HyperHier921	chr7	150147553	150148073	0.78	6.21	53145	47507	47514	8	9	1.75E-04	1.10E-02	GIMAP8 (+95)
HyperHier922	chr1	153762201	153762564	0.77	4.65	3841	6612	6617	6	6	5.07E-04	1.70E-02	SLC27A3 (+14615), GATAD2B (+133068)
HyperHier923	chr21	43823809	43824109	0.77	3.87	36457	102628	102632	5	6	1.01E-03	3.00E-02	UBASH3A (-65)
HyperHier924	chr16	1148030	1148520	0.77	3.09	21144	82990	82993	4	5	2.27E-03	5.90E-02	C1QTNF8 (-2031)
HyperHier925	chr17	80273242	80273685	0.77	3.08	26702	92691	92694	4	4	2.28E-03	5.90E-02	CSNK1D (-41857), CD7 (+2014)
HyperHier926	chr6	106433792	106434169	0.77	3.08	49070	39925	39928	4	9	2.28E-03	5.90E-02	PREP (-583022), PRDM1 (-100214)
HyperHier927	chr2	242953754	242954430	0.77	3.08	34832	17840	17843	4	4	2.29E-03	5.90E-02	CXXC11 (+142211)

HyperHier928	chr17	40489513	40489785	0.77	3.84	25014	89673	89677	5	5	1.04E-03	3.20E-02	STAT5A (+50084), STAT3 (+50937)
HyperHier929	chr11	2292677	2292787	0.76	3.05	9149	61697	61700	4	24	2.33E-03	6.20E-02	ASCL2 (-550)
HyperHier930	chr17	45949677	45949878	0.76	3.81	25299	90175	90179	5	5	1.06E-03	3.50E-02	SP2 (-23738), SP6 (-21219)
HyperHier931	chr10	73848817	73849167	0.76	5.33	7090	57665	57671	7	9	2.98E-04	1.50E-02	SPOCK2 (-461)
HyperHier932	chr7	50132653	50133000	0.76	3.80	51550	44688	44692	5	7	1.07E-03	3.60E-02	C7orf72 (-2805), ZPBP (+33)
HyperHier933	chr7	139256124	139256620	0.76	4.56	52973	47221	47226	6	6	5.42E-04	2.40E-02	CLEC2L (+47770), HIPK2 (+221145)
HyperHier934	chr7	2138722	2139216	0.76	3.03	50581	42681	42684	4	4	2.38E-03	7.00E-02	MAD1L1 (+133909), ELFN1 (+411214)
HyperHier935	chr13	114814024	114814706	0.76	6.07	17100	75926	75933	8	8	1.88E-04	1.60E-02	GAS6 (-247325), RASA3 (+83721)
HyperHier936	chr2	241721504	241722113	0.76	3.03	34738	17664	17667	4	5	2.40E-03	7.10E-02	KIF1A (+37916), AQP12A (+90547)
HyperHier937	chr10	135090098	135090565	0.75	3.77	8806	60919	60923	5	5	1.10E-03	4.10E-02	ADAM8 (+40)
HyperHier938	chr19	51231557	51232007	0.75	3.77	29894	98601	98605	5	5	1.10E-03	4.10E-02	GPR32 (-41939), CLEC11A (+5196)
HyperHier939	chr17	62075103	62075409	0.75	3.01	25802	91156	91159	4	4	2.44E-03	7.40E-02	SCN4A (-24978), ICAM2 (+22738)
HyperHier940	chr7	1894209	1895060	0.75	3.76	50550	42614	42618	5	5	1.12E-03	4.40E-02	ELFN1 (+166880), MAD1L1 (+378243)
HyperHier941	chr11	1082797	1083509	0.75	4.48	8969	61269	61274	6	6	5.76E-04	3.20E-02	MUC5AC (-68427), MUC2 (+8278)
HyperHier942	chr12	54763081	54763433	0.75	2.99	13793	69865	69868	4	4	2.51E-03	8.00E-02	GPR84 (-4986)
HyperHier943	chr6	146919932	146920295	0.75	6.72	49657	40986	40994	9	9	1.29E-04	1.40E-02	ADGB (+13)
HyperHier944	chr10	135342560	135343248	0.75	2.98	8836	60987	60990	4	8	2.52E-03	8.20E-02	CYP2E1 (+8994), SYCE1 (+36234)
HyperHier945	chr19	54927717	54928185	0.74	2.97	30112	99049	99052	4	6	2.55E-03	8.30E-02	LENG8 (-32114), TTYH1 (+1251)
HyperHier946	chr12	53183930	53184086	0.74	2.97	13641	69458	69461	4	5	2.56E-03	8.30E-02	KRT76 (-12879), KRT3 (+5893)
HyperHier947	chr7	27143478	27143788	0.74	2.97	51035	43656	43659	4	28	2.57E-03	8.30E-02	HOXA2 (-1203)
HyperHier948	chr6	34203153	34203887	0.74	2.96	48066	38081	38084	4	4	2.58E-03	8.40E-02	HMGA1 (-1130)
HyperHier949	chr1	45189875	45190912	0.74	3.71	2364	3916	3920	5	6	1.18E-03	4.70E-02	TMEM53 (-50167), KIF2C (-15098)
HyperHier950	chr16	85932383	85932853	0.74	2.96	23130	86274	86277	4	8	2.58E-03	8.50E-02	IRF8 (+209)
HyperHier951	chr22	24823110	24823554	0.74	4.44	36882	103326	103331	6	6	5.96E-04	3.40E-02	ADORA2A (-198)
HyperHier952	chr16	89005932	89006877	0.74	3.70	23430	86802	86806	5	5	1.18E-03	4.80E-02	PABPN1L (-73379), CBFA2T3 (+37207)
HyperHier953	chr15	68497992	68498857	0.74	2.94	20058	81004	81007	4	4	2.63E-03	9.60E-02	CALML4 (-526)
HyperHier954	chr17	72357992	72358370	0.74	3.68	26026	91471	91475	5	5	1.21E-03	5.50E-02	BTBD17 (-96)
HyperHier955	chr3	128779277	128779601	0.73	5.13	39691	21270	21276	7	7	3.44E-04	3.10E-02	GP9 (-171)
HyperHier956	chr13	107028903	107029443	0.73	3.66	16731	75213	75217	5	5	1.24E-03	5.70E-02	EFNB2 (+158289), DAOA (+910581)
HyperHier957	chr6	32014300	32016247	0.73	10.94	47806	37187	37201	15	21	1.42E-05	1.00E-03	TNXB (-1369)
HyperHier958	chr2	37571677	37571901	0.73	3.64	31203	11424	11428	5	5	1.26E-03	6.10E-02	QPCT (+72)
HyperHier959	chr3	52528955	52529524	0.73	6.54	38603	19559	19567	9	10	1.41E-04	1.80E-02	STAB1 (-114)
HyperHier960	chr4	1304836	1305425	0.73	3.63	40911	23522	23526	5	5	1.29E-03	6.30E-02	UVSSA (-35973), MAEA (+21492)
HyperHier961	chr3	52813652	52813920	0.72	3.59	38613	19582	19586	5	5	1.33E-03	7.70E-02	ITIH3 (-15028), ITIH1 (+2178)
HyperHier962	chr10	3281803	3282650	0.71	3.57	6034	55813	55817	5	5	1.36E-03	7.90E-02	PITRM1 (-67224), KLF6 (+545240)
HyperHier963	chr17	43922122	43922356	0.71	3.56	25238	90075	90079	5	6	1.38E-03	8.50E-02	SPPL2C (-17)
HyperHier964	chr5	154026371	154027256	0.71	4.27	45942	32928	32933	6	6	6.99E-04	5.90E-02	HAND1 (-168990), LARP1 (-65648)
HyperHier965	chr10	135123006	135123764	0.71	4.26	8811	60929	60934	6	6	7.03E-04	6.00E-02	ZNF511 (+1406), CALY (+27026)
HyperHier966	chr17	65471303	65471507	0.71	3.54	25886	91276	91280	5	5	1.41E-03	9.30E-02	NOL11 (-242544), PITPNC1 (+97134)
HyperHier967	chr6	31590513	31590870	0.71	3.53	47745	36947	36951	5	5	1.41E-03	9.40E-02	PRRC2A (+2190), BAG6 (+29785)
HyperHier968	chr1	156357871	156358408	0.71	4.23	3987	6870	6875	6	7	7.23E-04	6.70E-02	RHBG (+19137), C1orf61 (+41044)
HyperHier969	chr8	38757532	38757859	0.70	3.52	54653	50250	50254	5	5	1.43E-03	9.50E-02	PLEKHA2 (-1057)
HyperHier970	chr17	1477843	1479213	0.70	5.63	23688	87252	87259	8	8	2.49E-04	4.20E-02	PITPNA (-12418), SLC43A2 (+53602)
HyperHier971	chr12	123380870	123381003	0.70	3.52	15108	72241	72245	5	5	1.43E-03	9.70E-02	VPS37B (+54)
HyperHier972	chr6	31627617	31627714	0.70	4.21	47750	36961	36966	6	6	7.37E-04	7.00E-02	C6orf47 (+883)
HyperHier973	chr6	32121433	32121566	0.69	4.16	47845	37453	37458	6	18	7.68E-04	8.00E-02	PRRT1 (-1771), PPT2-EGFL8 (-499), PPT2 (+200)
HyperHier974	chr13	80054976	80055241	0.69	4.84	16456	74659	74665	7	7	4.32E-04	6.20E-02	NDFIP2 (-178)
HyperHier975	chr16	875257	876216	0.69	4.84	21077	82862	82868	7	7	4.32E-04	6.20E-02	GNG13 (-25004), LMF1 (+145262)
HyperHier976	chr6	3848790	3849190	0.69	4.80	46826	34456	34462	7	7	4.47E-04	7.10E-02	FAM50B (-630)
HyperHier977	chr11	70516627	70517411	0.67	4.72	11034	65231	65237	7	7	4.77E-04	9.10E-02	CTTN (+272372), SHANK2 (+341353)
HyperHier978	chr6	30881658	30881842	0.63	6.34	47669	36698	36707	10	19	1.61E-04	6.50E-02	VARS2 (-358)
HyperHier979	chr6	30039130	30039801	0.53	11.06	47565	36250	36270	21	24	1.32E-05	2.00E-02	RNF39 (+4198), PPP1R11 (+4601)
HypoHier662	chr6	101846767	101847706	-0.50	8.01	49034	39840	39855	16	19	6.53E-05	7.20E-02	GRIK2 (+573)
HypoHier661	chr1	3566950	3568243	-0.50	11.54	440	827	849	23	24	1.14E-05	7.00E-03	TP73 (-1487), WRAP73 (-960)
HypoHier660	chr2	176971820	176974060	-0.50	8.06	33427	15295	15310	16	16	6.34E-05	6.70E-02	HOXD10 (-8367), HOXD11 (+926)
HypoHier659	chr5	122429178	122431419	-0.52	7.76	45061	31112	31126	15	15	7.31E-05	6.30E-02	PRDM6 (+5483), CEP120 (+328698)
HypoHier658	chr7	27280914	27282444	-0.53	11.62	51091	43893	43914	22	23	1.11E-05	1.20E-02	EVX1 (-485)
HypoHier657	chr12	114845868	114847641	-0.54	8.66	14822	71620	71635	16	16	4.61E-05	4.00E-02	TBX5 (-508)

HypoHier656	chr3	62354546	62357176	-0.54	7.59	38813	19908	19921	14	14	7.92E-05	8.00E-02	FEZF2 (+3329), PTPRG (+808276)
HypoHier655	chr6	28557080	28558006	-0.55	9.34	47419	35683	35699	17	20	2.86E-05	3.10E-02	SCAND3 (-2431)
HypoHier654	chr5	63255045	63257710	-0.55	7.15	44350	29760	29772	13	19	1.02E-04	9.70E-02	HTR1A (+1402)
HypoHier653	chr4	174450016	174451954	-0.55	7.15	43012	27360	27372	13	25	1.02E-04	9.70E-02	HAND2 (+395)
HypoHier652	chr5	50678451	50679681	-0.56	7.28	44208	29525	29537	13	15	9.55E-05	8.30E-02	ISL1 (+145)
HypoHier651	chr7	45960367	45961778	-0.56	9.54	51481	44559	44575	17	22	2.67E-05	2.60E-02	IGFBP3 (-223)
HypoHier650	chr13	112720171	112721950	-0.56	7.87	16919	75548	75561	14	14	6.93E-05	5.40E-02	SOX1 (-852)
HypoHier649	chr3	62363642	62365402	-0.56	7.34	38816	19945	19957	13	18	9.20E-05	7.70E-02	FEZF2 (-5332), CADPS (+496532)
HypoHier648	chr1	91190005	91192803	-0.57	13.04	3101	5281	5303	23	23	5.44E-06	2.00E-03	BARHL2 (-8610), ZNF644 (+295626)
HypoHier647	chr1	217307696	217311511	-0.57	11.36	5193	8873	8892	20	27	1.21E-05	1.90E-02	ESRRG (-412809), GPATCH2 (+494820)
HypoHier646	chr8	55370407	55372569	-0.57	7.97	54896	50721	50734	14	18	6.62E-05	4.80E-02	SOX17 (+993)
HypoHier645	chr8	50822168	50823541	-0.57	6.84	54836	50597	50608	12	12	1.20E-04	9.30E-02	SNTG1 (-1378)
HypoHier644	chr5	170734856	170736572	-0.57	8.58	46183	33318	33332	15	15	4.75E-05	2.50E-02	TLX3 (-574)
HypoHier643	chr6	117197952	117199067	-0.57	6.88	49278	40276	40287	12	12	1.18E-04	9.10E-02	RFX6 (+87)
HypoHier642	chr2	176986659	176988284	-0.58	6.91	33435	15346	15357	12	24	1.17E-04	8.90E-02	HOXD9 (+384)
HypoHier641	chr6	50817193	50819236	-0.58	6.91	48554	38863	38874	12	13	1.17E-04	8.90E-02	TFAP2B (+31779)
HypoHier640	chr4	183062004	183065224	-0.58	8.64	43089	27502	27516	15	18	4.68E-05	2.20E-02	TENM3 (-100968)
HypoHier639	chr4	185939458	185941601	-0.58	7.52	43170	27640	27652	13	13	8.28E-05	6.20E-02	HELT (+447)
HypoHier638	chr2	154333568	154335698	-0.58	6.96	33097	14582	14593	12	18	1.14E-04	8.30E-02	RPRM (+689)
HypoHier637	chr12	45443808	45445385	-0.58	8.15	13370	69012	69025	14	15	6.10E-05	3.60E-02	DBX2 (+285)
HypoHier636	chr6	56111812	56112696	-0.58	7.59	48628	38978	38990	13	13	7.90E-05	6.00E-02	COL21A1 (+290)
HypoHier635	chr6	100895050	100897445	-0.59	8.24	49022	39749	39762	14	16	5.79E-05	3.40E-02	MCHR2 (-454149), SIM1 (+16557)
HypoHier634	chr5	11903145	11904912	-0.59	8.24	43826	28866	28879	14	14	5.79E-05	3.40E-02	CTNND2 (+126)
HypoHier633	chr17	32483711	32484259	-0.59	6.50	24606	88922	88932	11	13	1.44E-04	9.90E-02	ASIC2 (-863979), CCL2 (-98319)
HypoHier632	chr10	134597884	134600357	-0.59	12.43	8696	60655	60675	21	35	7.09E-06	1.00E-02	NKX6-2 (+435)
HypoHier631	chr6	29974083	29975427	-0.60	17.28	47558	36207	36235	29	31	1.42E-06	0.00E+00	ZNRD1 (-54276), HLA-A (+65718)
HypoHier630	chr4	111531926	111533517	-0.60	6.01	42382	26203	26212	10	14	1.95E-04	9.10E-02	PITX2 (+11537), ENPEP (+135493)
HypoHier629	chr5	63801834	63802993	-0.60	7.84	44355	29793	29805	13	13	7.02E-05	4.60E-02	RGS7BP (+288)
HypoHier628	chr2	45170322	45172265	-0.60	6.64	31356	11703	11713	11	11	1.36E-04	7.90E-02	SIX3 (+2392), SIX2 (+65275)
HypoHier627	chr4	147560126	147562073	-0.60	6.04	42738	26824	26833	10	10	1.92E-04	8.60E-02	EDNRA (-840984), POU4F2 (+1055)
HypoHier626	chr2	228028601	228029790	-0.61	7.26	34240	16804	16815	12	13	9.67E-05	5.30E-02	COL4A4 (-367), COL4A3 (-85)
HypoHier625	chr15	74218468	74219307	-0.61	6.06	20221	81239	81248	10	10	1.89E-04	8.60E-02	LOXL1 (+89)
HypoHier624	chr4	155663225	155664311	-0.61	6.07	42858	27032	27041	10	14	1.87E-04	8.50E-02	LRAT (-1355)
HypoHier623	chr3	147125287	147126703	-0.61	12.16	39980	21834	21853	20	39	8.04E-06	1.00E-02	ZIC4 (-3924), ZIC1 (-1176)
HypoHier622	chr11	12029738	12031099	-0.61	6.10	9449	62282	62291	10	13	1.85E-04	8.30E-02	DKK3 (+210)
HypoHier621	chr6	85483055	85484863	-0.61	6.10	48849	39395	39404	10	13	1.84E-04	8.30E-02	NT5E (-675850), TBX18 (-9722)
HypoHier620	chr2	107502354	107504169	-0.61	7.33	32386	13389	13400	12	12	9.36E-05	5.00E-02	ST6GAL2 (-160)
HypoHier619	chr1	27709634	27709928	-0.61	7.35	1670	2764	2775	12	13	9.20E-05	5.00E-02	CD164L2 (+12)
HypoHier618	chr14	57275967	57279275	-0.61	11.04	17702	77094	77111	18	20	1.35E-05	1.10E-02	OTX2 (-434)
HypoHier617	chr6	100911526	100912168	-0.61	6.76	49026	39791	39801	11	12	1.27E-04	7.10E-02	SIM1 (+958)
HypoHier616	chr5	5139866	5140935	-0.62	5.54	43704	28690	28698	9	18	2.63E-04	9.50E-02	ADAMTS16 (-42)
HypoHier615	chr6	10415597	10416090	-0.62	6.77	46970	34721	34731	11	18	1.26E-04	7.10E-02	TFAP2A (-374)
HypoHier614	chr10	118896012	118898082	-0.62	8.63	8099	59513	59526	14	14	4.71E-05	2.20E-02	VAX1 (+520)
HypoHier613	chr5	113696863	113699683	-0.62	11.10	44967	30940	30957	18	19	1.25E-05	1.10E-02	KCNN2 (+1631), TRIM36 (+817583)
HypoHier612	chr7	156795356	156797833	-0.62	8.03	53418	48063	48075	13	15	6.50E-05	4.00E-02	MNX1 (+6750), NOM1 (+54178)
HypoHier611	chr1	165323692	165326345	-0.62	10.51	4272	7326	7342	17	17	1.68E-05	1.10E-02	LMX1A (+933)
HypoHier610	chr4	93224895	93227270	-0.62	8.03	42227	25927	25939	13	13	6.48E-05	4.00E-02	GRID2 (+533)
HypoHier609	chr12	106978920	106980319	-0.62	6.21	14587	71173	71182	10	10	1.75E-04	7.30E-02	RFX4 (-15295), POLR3B (+228184)
HypoHier608	chr4	104640489	104641548	-0.62	5.60	42321	26090	26098	9	10	2.55E-04	8.90E-02	TACR3 (-46)
HypoHier607	chr3	147140118	147141614	-0.62	7.47	39988	21895	21906	12	13	8.47E-05	4.30E-02	ZIC1 (+13695)
HypoHier606	chr19	50193437	50194662	-0.62	5.61	29819	98423	98431	9	13	2.52E-04	8.70E-02	CPT1C (-323)
HypoHier605	chr5	131592974	131593413	-0.63	5.65	45173	31312	31320	9	10	2.46E-04	8.40E-02	PDLIM4 (-170)
HypoHier604	chr8	65289751	65292321	-0.63	11.33	55023	50970	50987	18	19	1.21E-05	1.10E-02	BHLHE22 (-201778)
HypoHier603	chr12	47224649	47226255	-0.63	6.30	13395	69067	69076	10	10	1.63E-04	6.70E-02	PCED1B (-247934), SLC38A4 (-5672)
HypoHier602	chr6	30227800	30228083	-0.63	5.67	47600	36449	36457	9	12	2.42E-04	8.20E-02	TRIM39 (-66679), TRIM26 (-46799)
HypoHier601	chr2	63275509	63276565	-0.63	5.67	31593	12064	12072	9	27	2.42E-04	8.20E-02	OTX1 (-1900)
HypoHier600	chr8	25901335	25902611	-0.63	5.68	54374	49792	49800	9	10	2.41E-04	8.20E-02	EBF2 (+940)
HypoHier599	chr16	77468277	77469540	-0.63	5.69	22868	85845	85853	9	12	2.40E-04	8.20E-02	ADAMTS18 (+102)

HypoHier598	chr5	1874703	1876719	-0.63	8.24	43552	28338	28350	13	13	5.79E-05	3.30E-02	IRX4 (+11621), NDUFS6 (+74197)
HypoHier597	chr6	123317124	123318023	-0.64	6.35	49338	40395	40404	10	10	1.60E-04	6.30E-02	CLVS2 (+458)
HypoHier596	chr2	176963948	176964720	-0.64	6.36	33423	15274	15283	10	13	1.59E-04	6.30E-02	HOXD10 (-16973), HOXD11 (-7680), HOXD12 (-124)
HypoHier595	chr19	9608637	9609704	-0.64	6.36	28248	95539	95548	10	10	1.59E-04	6.30E-02	ZNF560 (+112)
HypoHier594	chr3	147110322	147113092	-0.64	12.11	39974	21789	21807	19	25	8.75E-06	9.00E-03	PLSCR1 (-849056), ZIC4 (+10364)
HypoHier593	chr14	38052740	38055252	-0.64	6.38	17481	76711	76720	10	10	1.56E-04	6.10E-02	FOXA1 (+10243), MIPOL1 (+386841)
HypoHier592	chr11	20181456	20182122	-0.64	5.76	9687	62734	62742	9	16	2.27E-04	6.90E-02	DBX1 (+81)
HypoHier591	chr5	150326069	150326642	-0.64	5.77	45869	32795	32803	9	10	2.25E-04	6.90E-02	GPX3 (-73768), ZNF300 (-41811)
HypoHier590	chr2	14772312	14773457	-0.64	7.08	30771	10731	10741	11	11	1.07E-04	5.10E-02	NBAS (+928569)
HypoHier589	chr13	108518419	108519880	-0.64	5.79	16758	75255	75263	9	12	2.23E-04	6.70E-02	FAM155A (-67)
HypoHier588	chr6	28457210	28458328	-0.64	5.15	47395	35607	35614	8	8	3.38E-04	9.80E-02	ZSCAN23 (-46525), GPX6 (+25801)
HypoHier587	chr14	102025815	102027797	-0.64	12.89	18723	78784	78803	20	20	6.38E-06	6.00E-03	DIO3 (-882)
HypoHier586	chr6	100916916	100917871	-0.64	5.16	49028	39818	39825	8	8	3.37E-04	9.80E-02	SIM1 (-4589)
HypoHier585	chr12	128750971	128752586	-0.65	5.82	15267	72498	72506	9	9	2.17E-04	6.70E-02	TMEM132C (-169)
HypoHier584	chr7	121939827	121940992	-0.65	5.18	52642	46622	46629	8	8	3.30E-04	9.70E-02	AASS (-166534), FEZF1 (+4149)
HypoHier583	chr11	65816463	65816985	-0.65	5.83	10724	64619	64627	9	9	2.16E-04	6.70E-02	SF3B2 (-3083), GAL3ST3 (-73)
HypoHier582	chr6	84417933	84419360	-0.65	9.08	48830	39331	39344	14	15	3.52E-05	1.60E-02	SNAP91 (+480)
HypoHier581	chr6	32119616	32120203	-0.65	6.50	47844	37431	37440	10	10	1.45E-04	5.50E-02	PPT2-EGFL8 (-2089), PPT2 (-1390), PRRT1 (-181)
HypoHier580	chr7	93519220	93520527	-0.65	8.45	52116	45629	45641	13	14	5.06E-05	2.60E-02	TFPI2 (+429)
HypoHier579	chr8	17270347	17271322	-0.65	6.50	54135	49344	49353	10	10	1.45E-04	5.50E-02	MTMR7 (+1)
HypoHier578	chr4	66535145	66535655	-0.65	5.22	41933	25461	25468	8	8	3.23E-04	9.20E-02	EPHA5 (+661)
HypoHier577	chr6	133561649	133562776	-0.65	22.84	49475	40615	40649	35	39	4.73E-07	0.00E+00	EYA4 (-276)
HypoHier576	chr4	111541965	111543401	-0.65	5.22	42387	26238	26245	8	8	3.22E-04	9.20E-02	PITX2 (+1576), ENPEP (+145454)
HypoHier575	chr6	28174875	28176230	-0.65	5.88	47381	35567	35575	9	9	2.11E-04	6.20E-02	ZSCAN9 (-17500), ZKSCAN8 (+65837)
HypoHier574	chr2	183730940	183732062	-0.65	5.24	33551	15645	15652	8	8	3.20E-04	9.20E-02	FRZB (+389)
HypoHier573	chr4	122301573	122302268	-0.66	5.90	42512	26488	26496	9	11	2.09E-04	6.10E-02	QRFPR (+293)
HypoHier572	chr1	91182128	91183051	-0.66	7.21	3098	5254	5264	11	11	9.86E-05	4.40E-02	BARHL2 (+204)
HypoHier571	chr11	91957448	91958799	-0.66	5.25	11456	65883	65890	8	8	3.17E-04	9.00E-02	FAT3 (-127138)
HypoHier570	chr14	37051417	37051949	-0.66	5.26	17454	76610	76617	8	8	3.15E-04	8.70E-02	NKX2-8 (+129)
HypoHier569	chr3	192444985	192445594	-0.66	7.89	40641	23002	23013	12	12	6.88E-05	2.80E-02	HRASLS (-513624), FGF12 (-318452)
HypoHier568	chr11	71954982	71955599	-0.66	6.58	11093	65332	65341	10	10	1.40E-04	4.90E-02	PHOX2A (-71)
HypoHier567	chr1	107682302	107684425	-0.66	5.27	3261	5599	5606	8	9	3.10E-04	8.40E-02	NTNG1 (-78)
HypoHier566	chr6	29943188	29944093	-0.66	7.90	47555	36190	36201	12	13	6.83E-05	2.80E-02	ZNRD1 (-85390), HLA-A (+34604)
HypoHier565	chr2	124782117	124783254	-0.66	6.59	32686	13967	13976	10	11	1.39E-04	4.90E-02	CNTNAP5 (-178)
HypoHier564	chr5	168727752	168729267	-0.66	7.91	46152	33260	33271	12	12	6.81E-05	2.80E-02	SLIT3 (-377)
HypoHier563	chr8	145925258	145926089	-0.66	5.28	56389	53325	53332	8	9	3.06E-04	8.20E-02	ARHGAP39 (-94590), ZNF251 (+55316)
HypoHier562	chr8	85096868	85097429	-0.66	5.28	55290	51489	51496	8	9	3.06E-04	8.20E-02	RALYL (+39)
HypoHier561	chr7	116962516	116963502	-0.66	5.29	52588	46515	46522	8	9	3.05E-04	8.20E-02	WNT2 (+334)
HypoHier560	chr1	57887655	57889604	-0.66	7.27	2697	4515	4525	11	11	9.62E-05	4.20E-02	C8B (-456942), DAB1 (+1099)
HypoHier559	chr17	47209660	47210599	-0.66	7.94	25423	90515	90526	12	12	6.69E-05	2.70E-02	B4GALNT2 (-199)
HypoHier558	chr10	50887212	50887632	-0.66	5.30	6796	57159	57166	8	15	3.04E-04	8.10E-02	CHAT (+65339), OGDHL (+82947)
HypoHier557	chr3	192231892	192233698	-0.66	5.30	40637	22989	22996	8	8	3.03E-04	8.10E-02	HRASLS (-726119), FGF12 (-105957)
HypoHier556	chr10	106399877	106401517	-0.66	8.63	7901	59156	59168	13	13	4.75E-05	2.40E-02	SORCS3 (-162)
HypoHier555	chr12	22093960	22095330	-0.66	9.29	13136	68605	68618	14	15	3.03E-05	1.20E-02	CMAS (-104463), ABCC9 (-5037)
HypoHier554	chr11	7694814	7695528	-0.66	5.32	9339	62119	62126	8	8	2.99E-04	7.70E-02	CYB5R2 (+268)
HypoHier553	chr10	42862876	42863550	-0.67	5.32	6624	56827	56834	8	9	2.99E-04	7.60E-02	ZNF33B (+270779)
HypoHier552	chr6	29759947	29760909	-0.67	7.99	47544	36131	36142	12	13	6.57E-05	2.70E-02	HLA-G (-34328), HLA-F (+69243)
HypoHier551	chr2	189156549	189157566	-0.67	6.00	33576	15694	15702	9	9	1.96E-04	5.00E-02	GULP1 (-501)
HypoHier550	chr6	30094980	30095802	-0.67	15.99	47578	36345	36368	24	24	1.66E-06	0.00E+00	TRIM31 (-14508), TRIM40 (-9396)
HypoHier549	chr1	241520286	241520926	-0.67	4.68	5747	9827	9833	7	8	4.94E-04	9.50E-02	RGS7 (-76)
HypoHier548	chr2	98703324	98703698	-0.67	5.36	32203	13098	13105	8	10	2.94E-04	7.10E-02	CNGA3 (-259107), TMEM131 (-91157)
HypoHier547	chr4	46390832	46392657	-0.67	12.74	41784	25130	25148	19	19	6.62E-06	5.00E-03	GABRG1 (-265647), GABRA2 (+85502)
HypoHier546	chr8	31496353	31498345	-0.67	7.37	54511	50029	50039	11	11	9.08E-05	3.70E-02	NRG1 (-908379), WRN (+606032)
HypoHier545	chr6	28554680	28555653	-0.67	12.07	47418	35662	35679	18	22	8.75E-06	5.00E-03	SCAND3 (-55)
HypoHier544	chr7	94537805	94538408	-0.67	8.05	52132	45698	45709	12	12	6.36E-05	2.50E-02	PPP1R9A (+1158), PON1 (+415912)
HypoHier543	chr6	72130432	72130799	-0.67	6.04	48692	39092	39100	9	10	1.92E-04	4.70E-02	RIMS1 (-466111), OGFRL1 (+132110)
HypoHier542	chr11	101918140	101918575	-0.67	5.38	11596	66117	66124	8	11	2.92E-04	7.00E-02	ANGPTL5 (-131105), YAP1 (-62834)
HypoHier541	chr4	155664941	155665832	-0.67	6.06	42859	27042	27050	9	9	1.89E-04	4.50E-02	LRAT (+264)

HypoHier540	chr4	76555547	76556042	-0.67	7.41	42041	25626	25636	11	11	8.80E-05	3.50E-02	CDKL2 (+105)
HypoHier539	chr6	10384975	10386160	-0.67	6.07	46955	34672	34680	9	9	1.87E-04	4.50E-02	OFCC1 (-446016), TFAP2A (+29902)
HypoHier538	chr19	51170356	51172144	-0.67	6.07	29882	98566	98574	9	9	1.87E-04	4.50E-02	SYT3 (-29948), SHANK1 (+48945)
HypoHier537	chr20	36148320	36150061	-0.67	22.95	35405	100624	100657	34	36	4.73E-07	0.00E+00	NNAT (-426)
HypoHier536	chr7	8480779	8483710	-0.68	10.13	50811	43142	43156	15	16	2.08E-05	6.00E-03	NXPH1 (+8660)
HypoHier535	chr2	176946790	176949017	-0.68	11.48	33416	15234	15250	17	17	1.18E-05	5.00E-03	HOXD10 (-33403), HOXD11 (-24110), HOXD12 (-16554), HOXD13 (-9715), EVX2 (+737)
HypoHier534	chr3	145878431	145879710	-0.68	6.77	39950	21712	21721	10	10	1.26E-04	4.00E-02	PLOD2 (-117)
HypoHier533	chr3	170746029	170746586	-0.68	4.74	40285	22411	22417	7	7	4.70E-04	8.50E-02	SLC2A2 (-1769)
HypoHier532	chr10	99789825	99790910	-0.68	5.43	7620	58535	58542	8	10	2.81E-04	6.30E-02	CRTAC1 (+217)
HypoHier531	chr7	95025611	95025924	-0.68	5.44	52138	45721	45728	8	20	2.80E-04	6.20E-02	PON3 (-95)
HypoHier530	chr7	121956502	121957254	-0.68	4.76	52649	46670	46676	7	7	4.65E-04	8.20E-02	FEZF1 (-12319), RNF133 (+382332)
HypoHier529	chr14	60973462	60974461	-0.68	6.12	17762	77209	77217	9	11	1.82E-04	4.10E-02	SIX6 (-1707)
HypoHier528	chr15	68120393	68122574	-0.68	6.13	20043	80979	80987	9	9	1.80E-04	4.00E-02	PIAS1 (-225033), SKOR1 (+9442)
HypoHier527	chr2	237086398	237087777	-0.68	4.77	34518	17280	17286	7	7	4.57E-04	7.90E-02	GBX2 (-10076), ASB18 (+85900)
HypoHier526	chr1	190447232	190448126	-0.68	4.10	4624	7939	7944	6	7	8.14E-04	9.80E-02	FAM5C (-920)
HypoHier525	chr14	63511872	63512760	-0.68	4.10	17826	77353	77358	6	6	8.12E-04	9.70E-02	KCNH5 (-143)
HypoHier524	chr2	45155201	45156207	-0.68	5.47	31346	11659	11666	8	12	2.74E-04	5.80E-02	SIX3 (-13198), CAMKMT (+566601)
HypoHier523	chr10	8095797	8097331	-0.69	9.59	6153	56023	56036	14	18	2.62E-05	1.10E-02	GATA3 (-92)
HypoHier522	chr4	48987791	48988321	-0.69	5.48	41815	25213	25220	8	9	2.71E-04	5.40E-02	CWH43 (-208)
HypoHier521	chr1	228651848	228652581	-0.69	4.12	5468	9355	9360	6	9	8.01E-04	9.00E-02	RNF187 (-22547), HIST3H2BB (+6407)
HypoHier520	chr14	29242823	29244278	-0.69	6.18	17359	76411	76419	9	9	1.76E-04	3.30E-02	FOXG1 (+8501)
HypoHier519	chr3	6903921	6905031	-0.69	4.12	37724	17963	17968	6	6	8.00E-04	9.00E-02	GRM7 (+1674)
HypoHier518	chr6	31691506	31692375	-0.69	13.75	47765	37029	37048	20	28	4.26E-06	3.00E-03	LY6G6C (-2319), C6orf25 (+780)
HypoHier517	chr16	68771035	68772225	-0.69	4.13	22693	85576	85581	6	10	7.92E-04	8.60E-02	CDH1 (+502)
HypoHier516	chr4	186456009	186457012	-0.69	6.20	43190	27685	27693	9	9	1.75E-04	3.20E-02	PDLIM3 (+151)
HypoHier515	chr4	122685675	122686564	-0.69	6.20	42518	26510	26518	9	15	1.75E-04	3.20E-02	TMEM155 (+220)
HypoHier514	chr6	168719954	168720679	-0.69	4.13	50162	41869	41874	6	7	7.89E-04	8.50E-02	DACT2 (+85)
HypoHier513	chr6	27512849	27513479	-0.69	4.13	47338	35467	35472	6	6	7.88E-04	8.50E-02	ZNF184 (-72267), HIST1H2BL (+262545)
HypoHier512	chr3	62860802	62861311	-0.69	5.53	38830	19973	19980	8	12	2.64E-04	4.90E-02	CADPS (-3)
HypoHier511	chr2	14773973	14775494	-0.69	6.91	30772	10742	10751	10	10	1.17E-04	2.90E-02	NBAS (+926720)
HypoHier510	chr6	70576168	70576871	-0.69	4.84	48672	39054	39060	7	7	4.33E-04	6.20E-02	COL19A1 (+57)
HypoHier509	chr2	108602860	108603466	-0.69	6.92	32391	13406	13415	10	11	1.17E-04	2.90E-02	SLC5A7 (+168)
HypoHier508	chr6	19691654	19692418	-0.69	4.16	47156	35066	35071	6	6	7.69E-04	8.00E-02	ID4 (-145581)
HypoHier507	chr1	54821910	54822503	-0.69	4.16	2613	4403	4408	6	7	7.69E-04	8.00E-02	SSBP3 (+49885), C1orf191 (+118497)
HypoHier506	chr3	147126763	147128157	-0.69	9.03	39980	21855	21867	13	39	3.69E-05	1.70E-02	ZIC1 (+289)
HypoHier505	chr2	177052486	177053568	-0.70	9.05	33463	15491	15503	13	13	3.67E-05	1.50E-02	HOXD1 (-280)
HypoHier504	chr1	47489195	47489776	-0.70	4.18	2444	4047	4052	6	6	7.55E-04	7.60E-02	CYP4X1 (+246)
HypoHier503	chr6	125283969	125284659	-0.70	5.58	49354	40421	40428	8	8	2.58E-04	4.70E-02	RNF217 (-20200)
HypoHier502	chr7	96631384	96633530	-0.70	8.38	52172	45791	45802	12	13	5.27E-05	1.60E-02	DLX6 (-2403)
HypoHier501	chr6	78172192	78174065	-0.70	7.68	48782	39237	39247	11	11	7.54E-05	2.20E-02	HTR1B (+361)
HypoHier500	chr5	54516372	54516879	-0.70	4.90	44250	29610	29616	7	7	4.12E-04	5.60E-02	MCIDAS (+6517), GPX8 (+60680)
HypoHier499	chr6	28641622	28642394	-0.70	7.70	47426	35766	35776	11	11	7.50E-05	2.20E-02	SCAND3 (-86896), TRIM27 (+249758)
HypoHier498	chr6	42927940	42928920	-0.70	18.90	48368	38521	38547	27	27	9.46E-07	0.00E+00	GNMT (-66)
HypoHier497	chr10	71389696	71390385	-0.70	4.91	7008	57520	57526	7	8	4.04E-04	5.40E-02	C10orf35 (+27)
HypoHier496	chr14	54413371	54413931	-0.70	3.51	17627	76958	76962	5	6	1.44E-03	9.80E-02	DDHD1 (-793835), BMP4 (+9878)
HypoHier495	chr12	103350170	103351443	-0.70	5.62	14515	71021	71028	8	9	2.50E-04	4.20E-02	ASCL1 (-657)
HypoHier494	chr18	67067721	67068730	-0.70	4.22	27330	93955	93960	6	7	7.32E-04	6.80E-02	DOK6 (-65)
HypoHier493	chr12	54320564	54322092	-0.70	5.63	13707	69563	69570	8	10	2.50E-04	4.20E-02	CALCOCO1 (-200104), HOXC13 (-11207)
HypoHier492	chr18	31803067	31803836	-0.70	4.93	27043	93418	93424	7	7	3.99E-04	5.20E-02	NOL4 (+63)
HypoHier491	chr14	29234671	29236898	-0.70	18.30	17354	76381	76406	26	26	9.46E-07	0.00E+00	FOXG1 (+735)
HypoHier490	chr19	58629719	58630089	-0.70	4.93	30317	99449	99455	7	10	3.99E-04	5.20E-02	ZSCAN18 (-111)
HypoHier489	chr8	98289880	98290510	-0.70	5.64	55477	51768	51775	8	8	2.49E-04	4.20E-02	TSPYL5 (-19)
HypoHier488	chr19	39465969	39466757	-0.70	4.93	29193	97320	97326	7	8	3.97E-04	5.00E-02	FBXO17 (+187)
HypoHier487	chr4	121992261	121993098	-0.70	3.52	42507	26478	26482	5	6	1.43E-03	9.50E-02	NDNF (+993)
HypoHier486	chr18	12254452	12254566	-0.71	4.24	26908	93098	93103	6	8	7.19E-04	6.70E-02	CIDEA (+191)
HypoHier485	chr17	9862752	9863061	-0.71	5.65	24120	88114	88121	8	8	2.45E-04	3.90E-02	RCVRN (-53969), GAS7 (+238961)
HypoHier484	chr6	32116591	32117079	-0.71	11.32	47842	37398	37413	16	30	1.21E-05	2.00E-03	PPT2 (-4465), PRRT1 (+2894)
HypoHier483	chr7	1276617	1278508	-0.71	8.50	50446	42401	42412	12	12	5.01E-05	1.50E-02	UNCX (+5020), MICALL2 (+221575)

HypoHier482	chr14	62583787	62584600	-0.71	4.96	17819	77340	77346	7	8	3.89E-04	4.60E-02	SYT16 (+121653), KCNH5 (+927979)
HypoHier481	chr12	72666976	72667707	-0.71	5.69	14147	70409	70416	8	8	2.40E-04	3.70E-02	TRHDE (+879)
HypoHier480	chr8	69242921	69244734	-0.71	7.12	55084	51102	51111	10	10	1.05E-04	2.10E-02	PREX2 (+379475)
HypoHier479	chr17	27313033	27313499	-0.71	3.56	24475	88710	88714	5	5	1.37E-03	8.20E-02	PHF12 (-34477), SEZ6 (+20050)
HypoHier478	chr2	80529621	80530948	-0.71	4.99	31998	12763	12769	7	8	3.81E-04	4.20E-02	LRRTM1 (+1589)
HypoHier477	chr8	24813653	24814393	-0.71	3.56	54346	49736	49740	5	5	1.37E-03	8.10E-02	DOCK5 (-228357), NEFM (+43498)
HypoHier476	chr8	93113829	93114437	-0.71	3.57	55370	51620	51624	5	5	1.35E-03	7.70E-02	RUNX1T1 (-6427), TRIQK (+864240)
HypoHier475	chr7	121942915	121947425	-0.72	17.90	52645	46633	46657	25	25	1.42E-06	0.00E+00	FEZF1 (-611)
HypoHier474	chr6	29893605	29894248	-0.72	5.01	47552	36174	36180	7	7	3.76E-04	4.00E-02	HLA-A (-15110), HLA-G (+99171)
HypoHier473	chr15	65689058	65689433	-0.72	4.30	19975	80865	80870	6	6	6.79E-04	5.40E-02	IGDCC3 (-18868), IGDCC4 (+26164)
HypoHier472	chr11	105480347	105481863	-0.72	10.75	11646	66186	66200	15	15	1.49E-05	2.00E-03	GRIA4 (+305)
HypoHier471	chr12	39299326	39299726	-0.72	3.58	13319	68894	68898	5	5	1.34E-03	7.70E-02	KIF21A (+537262), ALG10B (+589146)
HypoHier470	chr14	105944604	105945685	-0.72	4.30	19003	79304	79309	6	7	6.77E-04	5.40E-02	CRIP1 (-7509), CRIP2 (+5846)
HypoHier469	chr1	67772896	67773044	-0.72	3.59	2868	4827	4831	5	5	1.34E-03	7.70E-02	IL12RB2 (-77)
HypoHier468	chr6	96462814	96464135	-0.72	5.02	48934	39541	39547	7	7	3.73E-04	4.00E-02	FUT9 (-385)
HypoHier467	chr2	155554688	155555414	-0.72	3.59	33109	14624	14628	5	5	1.33E-03	7.70E-02	KCNJ3 (+240)
HypoHier466	chr3	48699637	48700498	-0.72	7.19	38452	19272	19281	10	10	1.00E-04	2.00E-02	CELSR3 (+280)
HypoHier465	chr13	39261165	39262298	-0.72	5.76	16058	74001	74008	8	8	2.28E-04	3.30E-02	FREM2 (+466)
HypoHier464	chr10	57389676	57391271	-0.72	5.05	6856	57269	57275	7	8	3.65E-04	3.70E-02	MTRNR2L5 (+31724), ZWINT (+730562)
HypoHier463	chr3	119421604	119422359	-0.72	5.77	39423	20832	20839	8	8	2.26E-04	3.10E-02	MAATS1 (+113)
HypoHier462	chr14	38066873	38068553	-0.72	5.06	17488	76733	76739	7	14	3.64E-04	3.70E-02	FOXA1 (-3474)
HypoHier461	chr10	133109867	133110731	-0.72	3.62	8573	60422	60426	5	5	1.30E-03	7.20E-02	PPP2R2D (-637656)
HypoHier460	chr13	112722719	112723581	-0.72	4.34	16920	75562	75567	6	6	6.53E-04	4.80E-02	SPACA7 (-307483), SOX1 (+1237)
HypoHier459	chr3	46607965	46608139	-0.72	3.62	38386	19136	19140	5	5	1.29E-03	6.50E-02	TDGF1 (-10675), RTP3 (+69071)
HypoHier458	chr5	72598865	72599503	-0.72	3.62	44452	30009	30013	5	6	1.29E-03	6.40E-02	TMEM174 (+130162), FOXD1 (+145168)
HypoHier457	chr18	5629371	5630428	-0.72	5.07	26843	92974	92980	7	7	3.58E-04	3.30E-02	EPB41L3 (-85659), TMEM200C (+266054)
HypoHier456	chr5	3596996	3597760	-0.73	3.63	43674	28623	28627	5	9	1.29E-03	6.30E-02	IRX1 (+1210)
HypoHier455	chr5	140261653	140262304	-0.73	5.08	45485	31935	31941	7	7	3.57E-04	3.30E-02	PCDHA13 (+125)
HypoHier454	chr1	216896852	216897284	-0.73	3.63	5183	8850	8854	5	5	1.28E-03	6.30E-02	ESRRG (-273)
HypoHier453	chr11	128693961	128694388	-0.73	4.36	12344	67323	67328	6	6	6.42E-04	4.40E-02	KCNJ1 (+18254), FLI1 (+130285)
HypoHier452	chr10	94179972	94180753	-0.73	4.36	7489	58276	58281	6	6	6.39E-04	4.30E-02	MARCH5 (+129443), IDE (+153470)
HypoHier451	chr6	39901897	39902348	-0.73	4.36	48253	38344	38349	6	8	6.39E-04	4.30E-02	MOCS1 (-6668), LRFN2 (+653081)
HypoHier450	chr3	6902337	6903327	-0.73	5.10	37723	17956	17962	7	7	3.53E-04	3.20E-02	GRM7 (+30)
HypoHier449	chr6	30652771	30653732	-0.73	8.74	47640	36575	36586	12	16	4.45E-05	7.00E-03	DHX16 (-12438), PPP1R18 (+2420)
HypoHier448	chr12	45268999	45270896	-0.73	18.20	13365	68983	69007	25	26	1.18E-06	0.00E+00	NELL2 (+932)
HypoHier447	chr6	28510281	28510532	-0.73	5.83	47409	35637	35644	8	9	2.17E-04	2.70E-02	GPX5 (+16705), SCAND3 (+44705)
HypoHier446	chr1	75590483	75591353	-0.73	3.65	2922	4923	4927	5	5	1.26E-03	6.10E-02	LHX8 (-3201)
HypoHier445	chr4	154711304	154711563	-0.73	4.38	42843	26988	26993	6	32	6.32E-04	4.20E-02	SFRP2 (-1162)
HypoHier444	chr5	95295740	95296502	-0.73	3.65	44802	30695	30699	5	5	1.25E-03	6.10E-02	GLRX (-137703), ELL2 (+1654)
HypoHier443	chr3	132756780	132757973	-0.73	8.04	39739	21356	21366	11	12	6.46E-05	1.20E-02	TMEM108 (+142)
HypoHier442	chr2	105483826	105484823	-0.73	4.40	32344	13323	13328	6	6	6.24E-04	4.00E-02	MRPS9 (-170116), POU3F3 (+12356)
HypoHier441	chr4	164253006	164253656	-0.73	3.66	42921	27162	27166	5	5	1.24E-03	5.70E-02	NPY1R (+683)
HypoHier440	chr5	80529067	80529340	-0.73	4.40	44634	30363	30368	6	6	6.17E-04	3.90E-02	CKMT2 (+61)
HypoHier439	chr12	85305829	85307152	-0.73	9.55	14271	70603	70615	13	18	2.67E-05	4.00E-03	SLC6A15 (+164)
HypoHier438	chr1	1475143	1475742	-0.74	5.15	128	256	262	7	7	3.40E-04	3.00E-02	TMEM240 (+294)
HypoHier437	chr13	112759355	112760313	-0.74	4.41	16930	75600	75605	6	16	6.13E-04	3.90E-02	SPACA7 (-270799), SOX1 (+37921)
HypoHier436	chr17	11501503	11502254	-0.74	8.83	24159	88181	88192	12	12	4.23E-05	6.00E-03	DNAH9 (+131)
HypoHier435	chr3	85008587	85008828	-0.74	2.95	39121	20423	20426	4	4	2.62E-03	9.30E-02	CADM2 (-766924)
HypoHier434	chr17	53342740	53343561	-0.74	4.43	25579	90785	90790	6	6	6.05E-04	3.60E-02	HLF (+778)
HypoHier433	chr12	52626814	52627438	-0.74	6.64	13606	69398	69406	9	10	1.36E-04	1.50E-02	KRT7 (+228)
HypoHier432	chr7	1279790	1280491	-0.74	3.69	50448	42415	42419	5	5	1.20E-03	4.90E-02	UNCX (+7598), MICALL2 (+218997)
HypoHier431	chr6	31760233	31760629	-0.74	4.44	47775	37075	37080	6	11	5.99E-04	3.50E-02	VWA7 (-15360), VARS (+3299)
HypoHier430	chr12	125002007	125003064	-0.74	4.44	15171	72344	72349	6	11	5.97E-04	3.40E-02	NCOR2 (-22738), SCARB1 (+345857)
HypoHier429	chr8	15397637	15398333	-0.74	3.70	54114	49310	49314	5	6	1.19E-03	4.80E-02	TUSC3 (+193)
HypoHier428	chr11	89867679	89867976	-0.74	4.44	11443	65865	65870	6	10	5.96E-04	3.40E-02	NAALAD2 (+131)
HypoHier427	chr2	468179	469105	-0.74	5.19	30368	10113	10119	7	10	3.30E-04	2.60E-02	FAM150B (-180346), TMEM18 (+208797)
HypoHier426	chr13	108520391	108520945	-0.74	3.71	16759	75264	75268	5	5	1.18E-03	4.70E-02	FAM155A (-1585)
HypoHier425	chr16	49316845	49317262	-0.74	3.71	22211	84700	84704	5	5	1.18E-03	4.70E-02	CBLN1 (-1312)

HypoHier424	chr14	27067372	27068354	-0.74	4.45	17343	76356	76361	6	6	5.92E-04	3.30E-02	NOVA1 (-903)
HypoHier423	chr6	166074870	166075755	-0.74	4.45	49994	41558	41563	6	6	5.92E-04	3.30E-02	PDE10A (+244)
HypoHier422	chr6	27258466	27258767	-0.74	3.71	47320	35423	35427	5	10	1.17E-03	4.70E-02	ZNF391 (-97880), PRSS16 (+43115)
HypoHier421	chr10	135050326	135051022	-0.74	14.85	8800	60880	60899	20	31	2.60E-06	1.00E-03	VENTX (-234)
HypoHier420	chr8	101661767	101662053	-0.74	5.94	55525	51875	51882	8	11	2.02E-04	2.20E-02	SNX31 (-17)
HypoHier419	chr2	42067938	42068648	-0.74	4.46	31268	11527	11532	6	6	5.89E-04	3.30E-02	PKDCC (-206867)
HypoHier418	chr15	31621629	31621843	-0.74	2.98	19300	79811	79814	4	4	2.55E-03	8.30E-02	KLF13 (+2678), OTUD7A (+325806)
HypoHier417	chr7	1265635	1267228	-0.74	4.47	50440	42369	42374	6	6	5.86E-04	3.30E-02	ZFAND2A (-66618), UNCX (-6111)
HypoHier416	chr11	43568922	43569555	-0.75	2.98	10009	63425	63428	4	4	2.53E-03	8.20E-02	HSD17B12 (-133020), API5 (+235700)
HypoHier415	chr1	58715499	58716377	-0.75	7.46	2712	4545	4554	10	10	8.49E-05	1.30E-02	DAB1 (-826209), OMA1 (+296534)
HypoHier414	chr6	27342438	27343671	-0.75	5.23	47328	35443	35449	7	7	3.21E-04	2.50E-02	ZNF391 (-13442), PRSS16 (+127553)
HypoHier413	chr2	115919785	115920221	-0.75	2.99	32515	13617	13620	4	8	2.52E-03	8.20E-02	DPP10 (+824)
HypoHier412	chr6	117584596	117584815	-0.75	2.99	49281	40290	40293	4	4	2.51E-03	8.00E-02	VGLL2 (-2031)
HypoHier411	chr3	26663551	26663885	-0.75	4.48	38072	18551	18556	6	13	5.75E-04	3.20E-02	LRRC3B (-579)
HypoHier410	chr1	6208717	6209266	-0.75	2.99	561	1033	1036	4	4	2.51E-03	8.00E-02	CHD5 (+31191), KCNAB2 (+103011)
HypoHier409	chr5	140305713	140306458	-0.75	8.23	45487	31943	31953	11	11	5.79E-05	1.10E-02	PCDHAC1 (-392)
HypoHier408	chr4	90757139	90757533	-0.75	4.49	42218	25913	25918	6	6	5.71E-04	3.10E-02	SNCA (+954)
HypoHier407	chr4	206232	206877	-0.75	4.50	40806	23291	23296	6	6	5.70E-04	3.10E-02	ZNF732 (+92555), ZNF595 (+153345)
HypoHier406	chr8	110987530	110988256	-0.75	6.00	55646	52092	52099	8	9	1.96E-04	2.00E-02	KCNV1 (+183)
HypoHier405	chr11	30037696	30039113	-0.75	12.77	9815	63012	63028	17	17	6.62E-06	0.00E+00	KCNA4 (+165)
HypoHier404	chr6	29691981	29692111	-0.75	5.26	47538	36110	36116	7	7	3.13E-04	2.30E-02	HLA-F (+861)
HypoHier403	chr2	1747700	1749076	-0.75	6.01	30447	10243	10250	8	8	1.95E-04	2.00E-02	PXDN (-110)
HypoHier402	chr4	111549880	111551139	-0.75	6.02	42392	26260	26267	8	8	1.93E-04	1.80E-02	PITX2 (-6251)
HypoHier401	chr7	70597058	70598282	-0.75	8.29	51802	45129	45139	11	11	5.70E-05	1.00E-02	WBSCR17 (+515)
HypoHier400	chr4	81106414	81107185	-0.76	5.30	42105	25718	25724	7	7	3.03E-04	2.00E-02	PRDM8 (+376)
HypoHier399	chr20	26188639	26189240	-0.76	5.30	35220	100321	100327	7	7	3.01E-04	1.90E-02	ZNF337 (-521347)
HypoHier398	chr6	31650735	31651283	-0.76	12.89	47759	36995	37011	17	17	6.38E-06	0.00E+00	LY6G5C (-2855)
HypoHier397	chr1	32930375	32931179	-0.76	7.61	1874	3097	3106	10	10	7.90E-05	8.00E-03	ZBTB8B (+107)
HypoHier396	chr8	1771973	1772393	-0.76	3.81	53795	48725	48729	5	5	1.07E-03	3.60E-02	ARHGEF10 (+41)
HypoHier395	chr3	170302890	170304031	-0.76	7.62	40271	22384	22393	10	12	7.87E-05	8.00E-03	SLC7A14 (+402)
HypoHier394	chr16	49499006	49499385	-0.76	3.05	22216	84709	84712	4	4	2.34E-03	6.50E-02	CBLN1 (-183454), ZNF423 (+357454)
HypoHier393	chr10	63422630	63423079	-0.76	4.57	6906	57353	57358	6	6	5.39E-04	2.40E-02	ARID5B (-238204), TMEM26 (-209647)
HypoHier392	chr5	3605783	3607134	-0.76	9.15	43681	28641	28652	12	12	3.24E-05	2.00E-03	IRX1 (+10291)
HypoHier391	chr1	237205098	237205409	-0.76	3.05	5691	9743	9746	4	4	2.33E-03	6.20E-02	RYR2 (-251)
HypoHier390	chr4	41875193	41875784	-0.77	3.06	41749	25047	25050	4	5	2.32E-03	6.10E-02	PHOX2B (-124502), TMEM33 (-61648)
HypoHier389	chr7	44185323	44186183	-0.77	3.06	51440	44490	44493	4	4	2.32E-03	6.10E-02	MYL7 (-4822)
HypoHier388	chr1	183154981	183155644	-0.77	6.13	4567	7852	7859	8	9	1.81E-04	1.30E-02	LAMC2 (-110)
HypoHier387	chr12	59313279	59314400	-0.77	7.67	13980	70169	70178	10	10	7.73E-05	8.00E-03	LRIG3 (+463)
HypoHier386	chr13	58203618	58204379	-0.77	5.37	16328	74422	74428	7	7	2.93E-04	1.50E-02	PCDH17 (-2656)
HypoHier385	chr8	22089400	22089778	-0.77	3.84	54246	49529	49533	5	6	1.04E-03	3.20E-02	POLR3D (-13159), BMP1 (+67340)
HypoHier384	chr8	27348650	27348940	-0.77	3.84	54414	49895	49899	5	8	1.04E-03	3.20E-02	EPHX2 (+499)
HypoHier383	chr4	188916496	188917251	-0.77	9.99	43279	27843	27855	13	13	2.22E-05	2.00E-03	ZFP42 (-51)
HypoHier382	chr6	33040535	33041343	-0.77	6.15	47938	37701	37708	8	10	1.79E-04	1.30E-02	HLA-DPB1 (-2764)
HypoHier381	chr18	55103353	55103785	-0.77	3.08	27234	93776	93779	4	4	2.28E-03	5.90E-02	ONECUT2 (+652)
HypoHier380	chr3	40428383	40429014	-0.77	6.16	38245	18862	18869	8	8	1.78E-04	1.20E-02	ENTPD3 (+52)
HypoHier379	chr2	237076164	237079122	-0.77	13.87	34513	17245	17262	18	26	3.78E-06	0.00E+00	GBX2 (-631)
HypoHier378	chr2	200468445	200469026	-0.77	3.86	33684	15865	15869	5	5	1.02E-03	3.10E-02	SATB2 (-145917), FTCDNL1 (+247130)
HypoHier377	chr3	55520834	55521529	-0.77	5.40	38684	19710	19716	7	7	2.86E-04	1.30E-02	LRTM1 (-559081), WNT5A (+2791)
HypoHier376	chr1	62660556	62660861	-0.77	3.09	2777	4663	4666	4	6	2.26E-03	5.90E-02	KANK4 (+124376), INADL (+452560)
HypoHier375	chr16	1429863	1430103	-0.77	3.09	21216	83116	83119	4	4	2.26E-03	5.90E-02	GNPTG (+28059), UNKL (+34709)
HypoHier374	chr3	64670459	64672609	-0.77	10.05	38882	20069	20081	13	13	2.18E-05	1.00E-03	PRICKLE2 (-460403), ADAMTS9 (+2142)
HypoHier373	chr2	121412005	121412432	-0.77	3.09	32642	13902	13905	4	4	2.25E-03	5.80E-02	GLI2 (-137766), INHBB (+308500)
HypoHier372	chr19	20278013	20278423	-0.77	4.64	28826	96600	96605	6	6	5.10E-04	1.70E-02	ZNF486 (+135)
HypoHier371	chr1	228075423	228075749	-0.77	3.10	5428	9241	9244	4	4	2.23E-03	5.80E-02	WNT9A (+60013), PRSS38 (+72192)
HypoHier370	chr6	126069089	126070091	-0.78	3.10	49371	40454	40457	4	4	2.23E-03	5.70E-02	HEY2 (-1136)
HypoHier369	chr19	3687751	3688477	-0.78	4.66	27921	94968	94973	6	6	5.01E-04	1.70E-02	CACTIN (-61301), PIP5K1C (+12363)
HypoHier368	chr18	28621490	28623159	-0.78	13.21	27015	93332	93348	17	17	5.44E-06	0.00E+00	DSC3 (+382)
HypoHier367	chr19	44952417	44952808	-0.78	5.44	29431	97729	97735	7	7	2.80E-04	1.20E-02	ZNF229 (+52)

HypoHier366	chr19	23945472	23945903	-0.78	3.89	28869	96691	96695	5	5	9.97E-04	3.00E-02	ZNF681 (-3995), RPSAP58 (-128)
HypoHier365	chr3	154145810	154146871	-0.78	6.22	40087	22043	22050	8	8	1.74E-04	1.10E-02	DHX36 (-104055), GPR149 (+1163)
HypoHier364	chr4	187644620	187647690	-0.78	15.56	43246	27787	27806	20	20	1.66E-06	0.00E+00	FAT1 (-1146)
HypoHier363	chr14	33403060	33403226	-0.78	3.11	17396	76481	76484	4	4	2.20E-03	5.70E-02	NPAS3 (-5380), AKAP6 (+604664)
HypoHier362	chr5	178986131	178986906	-0.78	7.01	46568	34011	34019	9	9	1.10E-04	7.00E-03	RUFY1 (+8960), HNRNPH1 (+65151)
HypoHier361	chr2	5831147	5831722	-0.78	3.12	30547	10399	10402	4	4	2.19E-03	5.60E-02	SOX11 (-1364)
HypoHier360	chr3	185000208	185001026	-0.78	5.46	40517	22790	22796	7	7	2.77E-04	1.20E-02	MAP3K13 (-80291), EHHADH (-28731)
HypoHier359	chr6	32976805	32977708	-0.78	6.24	47936	37688	37695	8	8	1.72E-04	1.10E-02	HLA-DOA (+132)
HypoHier358	chr6	28602543	28603779	-0.78	25.78	47424	35732	35764	33	33	2.36E-07	0.00E+00	SCAND3 (-48049), TRIM27 (+288605)
HypoHier357	chr10	125034002	125034818	-0.78	5.47	8302	59934	59940	7	8	2.73E-04	1.20E-02	GPR26 (-391461), BUB3 (+120617)
HypoHier356	chr6	26271193	26271827	-0.78	3.91	47262	35291	35295	5	5	9.73E-04	3.00E-02	HIST1H2BI (-1634), HIST1H3G (+102)
HypoHier355	chr13	28553501	28553805	-0.78	3.91	15876	73675	73679	5	5	9.72E-04	3.00E-02	CDX2 (-8377), URAD (+9121)
HypoHier354	chr1	170630070	170630734	-0.78	4.69	4360	7483	7488	6	6	4.89E-04	1.70E-02	PRRX1 (-2645)
HypoHier353	chr10	26222922	26223310	-0.78	3.13	6434	56539	56542	4	6	2.17E-03	5.30E-02	MYO3A (-80)
HypoHier352	chr10	128593624	128594417	-0.78	5.48	8428	60137	60143	7	7	2.72E-04	1.20E-02	DOCK1 (+43)
HypoHier351	chr12	110156245	110156459	-0.78	3.92	14669	71316	71320	5	5	9.64E-04	3.00E-02	MMAB (-144673), TRPV4 (+96344)
HypoHier350	chr3	12045449	12046420	-0.78	4.71	37849	18201	18206	6	8	4.83E-04	1.70E-02	TAMM41 (-157542), TIMP4 (+154916)
HypoHier349	chr19	12758716	12759156	-0.78	3.92	28405	95844	95848	5	5	9.61E-04	3.00E-02	MAN2B1 (+18620), ZNF791 (+37204)
HypoHier348	chr16	30907560	30908233	-0.78	3.14	22051	84446	84449	4	4	2.15E-03	5.20E-02	BCL7C (-1616), CTF1 (-31)
HypoHier347	chr4	82135607	82135987	-0.79	2.36	42122	25763	25765	3	3	5.22E-03	9.80E-02	PRKG2 (+391)
HypoHier346	chr19	58520847	58521273	-0.79	3.14	30308	99411	99414	4	4	2.14E-03	5.10E-02	ZSCAN1 (-24374), ZNF606 (-6343)
HypoHier345	chr7	12443529	12443926	-0.79	3.93	50830	43183	43187	5	7	9.56E-04	2.90E-02	VWDE (-197)
HypoHier344	chr22	36806001	36806580	-0.79	3.93	37100	103688	103692	5	5	9.56E-04	2.90E-02	MYH9 (-22228), TXN2 (+71786)
HypoHier343	chr5	1882775	1883413	-0.79	6.29	43557	28364	28371	8	8	1.65E-04	1.10E-02	IRX4 (+4238), NDUFS6 (+81580)
HypoHier342	chr14	103739856	103740597	-0.79	3.93	18807	78968	78972	5	5	9.55E-04	2.90E-02	EIF5 (-60112), TNFAIP2 (+150429)
HypoHier341	chr7	1282630	1283255	-0.79	2.36	50450	42422	42424	3	7	5.19E-03	9.60E-02	UNCX (+10400), MICALL2 (+216195)
HypoHier340	chr2	214148958	214149735	-0.79	2.36	33883	16167	16169	3	3	5.18E-03	9.60E-02	SPAG16 (+234)
HypoHier339	chr13	51417469	51417929	-0.79	3.15	16241	74292	74295	4	4	2.13E-03	5.00E-02	RNASEH2B (-66115), DLEU1 (+761392)
HypoHier338	chr4	115519436	115520072	-0.79	7.09	42454	26393	26401	9	10	1.06E-04	7.00E-03	UGT8 (+143)
HypoHier337	chr6	114177329	114177535	-0.79	3.15	49223	40199	40202	4	4	2.11E-03	5.00E-02	MARCKS (-1109)
HypoHier336	chr12	47219626	47219958	-0.79	8.68	13394	69056	69066	11	11	4.56E-05	5.00E-03	SLC38A4 (-12)
HypoHier335	chr13	43566633	43566902	-0.79	2.37	16106	74076	74078	3	3	5.13E-03	9.50E-02	DNAJC15 (-30571), TNFSF11 (+418479)
HypoHier334	chr6	24910562	24911615	-0.79	11.06	47219	35171	35184	14	14	1.35E-05	0.00E+00	FAM65B (+106)
HypoHier333	chr6	46138699	46139019	-0.79	6.32	48486	38705	38712	8	8	1.62E-04	1.00E-02	ENPP5 (-151)
HypoHier332	chr12	54426670	54428592	-0.79	11.86	13757	69775	69789	15	15	9.93E-06	0.00E+00	HOXC5 (+994)
HypoHier331	chr5	16179210	16180419	-0.79	12.66	43888	28955	28970	16	19	6.62E-06	0.00E+00	MARCH11 (+69)
HypoHier330	chr1	91194674	91195504	-0.79	3.96	3102	5304	5308	5	10	9.30E-04	2.70E-02	BARHL2 (-12295), ZNF644 (+291941)
HypoHier329	chr4	148402128	148402899	-0.79	4.75	42755	26854	26859	6	8	4.68E-04	1.50E-02	EDNRA (+430)
HypoHier328	chr7	94284258	94285911	-0.79	22.97	52128	45662	45690	29	32	4.73E-07	0.00E+00	PEG10 (-552), SGCE (+351)
HypoHier327	chr2	172972681	172973241	-0.79	3.17	33329	15068	15071	4	4	2.07E-03	4.90E-02	ITGA6 (-319556), DLX2 (-5333)
HypoHier326	chr3	64253534	64253818	-0.79	4.76	38867	20042	20047	6	6	4.65E-04	1.40E-02	PRICKLE2 (-42545), ADAMTS9 (+420000)
HypoHier325	chr18	35145353	35147826	-0.79	18.24	27092	93500	93522	23	23	1.18E-06	0.00E+00	CELF4 (-590)
HypoHier324	chr4	188953092	188953969	-0.79	6.35	43282	27860	27867	8	8	1.59E-04	9.00E-03	ZFP42 (+36606), TRIML2 (+73216)
HypoHier323	chr11	111169284	111169473	-0.79	3.18	11714	66304	66307	4	4	2.05E-03	4.80E-02	ARHGAP20 (-585928), C11orf92 (+6391)
HypoHier322	chr12	85673200	85673347	-0.80	3.18	14275	70630	70633	4	4	2.04E-03	4.80E-02	ALX1 (-611)
HypoHier321	chr19	52956622	52957180	-0.80	7.96	30010	98838	98847	10	11	6.64E-05	6.00E-03	ZNF578 (+72)
HypoHier320	chr13	53422381	53422871	-0.80	6.37	16279	74352	74359	8	12	1.58E-04	9.00E-03	PCDH8 (+149)
HypoHier319	chr6	134638742	134639355	-0.80	5.58	49507	40725	40731	7	9	2.58E-04	1.00E-02	SGK1 (+147)
HypoHier318	chr3	2139977	2141750	-0.80	7.17	37683	17887	17895	9	11	1.01E-04	7.00E-03	CNTN4 (-139649)
HypoHier317	chr17	17603531	17604184	-0.80	4.80	24284	88409	88414	6	6	4.47E-04	1.40E-02	RAI1 (+19071), SREBF1 (+136443)
HypoHier316	chr1	18956947	18957232	-0.80	3.20	1220	2002	2005	4	22	1.99E-03	4.70E-02	PAX7 (-410)
HypoHier315	chr19	11784344	11785248	-0.80	10.42	28383	95796	95808	13	14	1.75E-05	1.00E-03	ZNF823 (+64992), ZNF627 (+76561)
HypoHier314	chr13	93879303	93879769	-0.80	4.01	16504	74751	74755	5	5	8.83E-04	2.60E-02	GPC6 (+441)
HypoHier313	chr14	33402027	33402512	-0.80	3.21	17395	76477	76480	4	5	1.98E-03	4.60E-02	NPAS3 (-6253), AKAP6 (+603791)
HypoHier312	chr11	7272867	7274234	-0.80	8.03	9321	62088	62097	10	10	6.50E-05	6.00E-03	SYT9 (+370)
HypoHier311	chr6	152128535	152129400	-0.80	4.82	49761	41170	41175	6	10	4.40E-04	1.40E-02	ESR1 (+117337), SYNE1 (+829566)
HypoHier310	chr7	158110405	158110685	-0.80	2.41	53585	48384	48386	3	4	4.86E-03	8.30E-02	PTPRN2 (+269826), DNAJB6 (+980885)
HypoHier309	chr3	62304470	62305213	-0.80	6.44	38810	19893	19900	8	11	1.52E-04	8.00E-03	FEZF2 (+54348), PTPRG (+757257)

HypoHier308	chr16	82044738	82045222	-0.80	7.24	22929	85941	85949	9	9	9.67E-05	6.00E-03	SDR42E1 (+113)
HypoHier307	chr6	43211208	43211235	-0.80	2.41	48386	38570	38572	3	6	4.85E-03	8.20E-02	TTBK1 (-196)
HypoHier306	chr15	91499626	91500193	-0.80	2.41	20698	82133	82135	3	3	4.85E-03	8.20E-02	RCCD1 (+1810), PRC1 (+37815)
HypoHier305	chr11	7041196	7041987	-0.81	12.08	9313	62061	62075	15	15	8.75E-06	0.00E+00	NLRP14 (-85), ZNF214 (+7)
HypoHier304	chr6	27647843	27649120	-0.81	8.86	47356	35511	35521	11	11	4.09E-05	5.00E-03	ZNF184 (-207585), HIST1H2BL (+127227)
HypoHier303	chr11	58672888	58673615	-0.81	5.64	10347	63986	63992	7	7	2.47E-04	8.00E-03	GLYATL2 (-60995), GLYATL1 (-37470)
HypoHier302	chr18	72916776	72917387	-0.81	3.23	27360	94021	94024	4	7	1.95E-03	4.00E-02	ZADH2 (+4221), ZNF407 (+574106)
HypoHier301	chr1	228872121	228872246	-0.81	2.43	5480	9388	9390	3	3	4.79E-03	7.60E-02	TMEM78 (-513199), RHOU (+1360)
HypoHier300	chr2	19561035	19561709	-0.81	3.24	30840	10863	10866	4	4	1.93E-03	3.80E-02	OSR1 (-4695)
HypoHier299	chr3	44726891	44727195	-0.81	3.24	38322	19008	19011	4	4	1.92E-03	3.80E-02	ZNF502 (-27095), ZNF35 (+36824)
HypoHier298	chr19	23185714	23185968	-0.81	2.43	28861	96670	96672	3	3	4.74E-03	7.20E-02	ZNF728 (+137)
HypoHier297	chr19	22444509	22444714	-0.81	2.43	28842	96637	96639	3	3	4.74E-03	7.20E-02	ZNF676 (-64859), ZNF729 (-24598)
HypoHier296	chr1	3105151	3105252	-0.81	2.44	373	713	715	3	3	4.73E-03	7.10E-02	ARHGEF16 (-265788), PRDM16 (+119427)
HypoHier295	chr20	1927546	1927911	-0.81	3.25	34900	99632	99635	4	4	1.90E-03	3.70E-02	PDYN (+46974), SIRPA (+51787)
HypoHier294	chr20	26190328	26190354	-0.81	2.44	35221	100328	100330	3	3	4.72E-03	6.80E-02	ZNF337 (-522748)
HypoHier293	chr10	49813265	49813434	-0.81	4.88	6763	57070	57075	6	13	4.17E-04	8.00E-03	ARHGAP22 (-353)
HypoHier292	chr4	16085716	16086085	-0.81	4.07	41484	24599	24603	5	9	8.34E-04	1.60E-02	PROM1 (-577)
HypoHier291	chr3	52351355	52351963	-0.81	4.07	38598	19549	19553	5	5	8.32E-04	1.60E-02	DNAH1 (+1324), BAP1 (+92707)
HypoHier290	chr2	237072391	237072998	-0.82	4.08	34509	17236	17240	5	5	8.27E-04	1.60E-02	GBX2 (+4317), AGAP1 (+669944)
HypoHier289	chr19	52598653	52599329	-0.82	5.71	30002	98806	98812	7	7	2.36E-04	6.00E-03	ZNF841 (+27)
HypoHier288	chr19	52872916	52873624	-0.82	7.35	30007	98819	98827	9	9	9.20E-05	5.00E-03	ZNF880 (+66)
HypoHier287	chr6	25652381	25652407	-0.82	4.90	47233	35203	35208	6	10	4.10E-04	8.00E-03	SCGN (-70)
HypoHier286	chr8	24771114	24773268	-0.82	14.74	54339	49709	49726	18	18	2.60E-06	0.00E+00	DOCK5 (-270189), NEFM (+1666)
HypoHier285	chr6	1312475	1312577	-0.82	4.92	46702	34234	34239	6	6	4.03E-04	8.00E-03	FOXQ1 (-149)
HypoHier284	chr12	79257667	79258438	-0.82	8.20	14211	70492	70501	10	10	5.89E-05	4.00E-03	SYT1 (-396)
HypoHier283	chr10	52834498	52835078	-0.82	4.10	6822	57214	57218	5	5	8.13E-04	1.60E-02	PRKG1 (+854)
HypoHier282	chr12	85667353	85667616	-0.82	2.46	14273	70622	70624	3	3	4.60E-03	6.20E-02	TSPAN19 (-237450), ALX1 (-6400)
HypoHier281	chr7	54609587	54610320	-0.82	7.38	51603	44787	44795	9	9	9.06E-05	5.00E-03	VSTM2A (-64)
HypoHier280	chr14	37666810	37667261	-0.82	4.10	17479	76705	76709	5	5	8.10E-04	1.60E-02	MIPOL1 (-119)
HypoHier279	chr17	8702564	8702868	-0.82	3.29	24083	88033	88036	4	4	1.82E-03	3.30E-02	MFSD6L (-49)
HypoHier278	chr5	178421314	178422415	-0.82	6.58	46536	33954	33961	8	8	1.39E-04	6.00E-03	GRM6 (+259)
HypoHier277	chr6	32920102	32920784	-0.82	4.95	47928	37670	37675	6	6	3.92E-04	6.00E-03	HLA-DMA (+456), ENSG00000248993 (+456)
HypoHier276	chr7	82791675	82792345	-0.83	4.13	52011	45454	45458	5	5	7.92E-04	1.40E-02	PCLO (+236)
HypoHier275	chr7	121950282	121950975	-0.83	4.13	52646	46658	46662	5	5	7.92E-04	1.40E-02	FEZF1 (-6070), RNF133 (+388581)
HypoHier274	chr12	5019933	5020693	-0.83	2.48	12771	68017	68019	3	3	4.49E-03	5.90E-02	KCNA5 (-132772), KCNA1 (+1242)
HypoHier273	chr6	29911542	29911558	-0.83	2.48	47554	36186	36188	3	3	4.49E-03	5.90E-02	ZNRD1 (-117481), HLA-A (+2513)
HypoHier272	chr9	14313043	14313762	-0.83	2.48	56493	53498	53500	3	3	4.49E-03	5.90E-02	NFIB (+263)
HypoHier271	chr11	86085366	86086101	-0.83	5.79	11405	65792	65798	7	7	2.24E-04	4.00E-03	C11orf73 (+72469), ME3 (+297944)
HypoHier270	chr5	137224213	137224967	-0.83	4.14	45334	31669	31673	5	8	7.85E-04	1.30E-02	PKD2L2 (-568)
HypoHier269	chr5	1886828	1888033	-0.83	4.97	43560	28381	28386	6	6	3.86E-04	6.00E-03	IRX4 (-99)
HypoHier268	chr19	58446312	58446988	-0.83	9.12	30304	99396	99406	11	11	3.41E-05	3.00E-03	ZNF418 (+90)
HypoHier267	chr4	166793789	166796183	-0.83	12.43	42948	27227	27241	15	15	7.09E-06	0.00E+00	TLL1 (+576)
HypoHier266	chr3	147108512	147108916	-0.83	2.49	39973	21780	21782	3	3	4.44E-03	5.70E-02	PLSCR1 (-846063), ZIC4 (+13357)
HypoHier265	chr7	105221826	105222180	-0.83	6.64	52454	46328	46335	8	8	1.36E-04	3.00E-03	EFCAB10 (-60)
HypoHier264	chr19	53811704	53812098	-0.83	3.32	30034	98892	98895	4	4	1.76E-03	2.50E-02	ZNF845 (-25101), BIRC8 (-17026)
HypoHier263	chr7	149158185	149158486	-0.83	2.49	53106	47437	47439	3	3	4.41E-03	5.40E-02	ZNF777 (-122)
HypoHier262	chr10	89264080	89264128	-0.83	2.49	7411	58155	58157	3	3	4.41E-03	5.40E-02	MINPP1 (-528)
HypoHier261	chr16	54968727	54969146	-0.83	4.99	22350	84968	84973	6	6	3.80E-04	5.00E-03	IRX6 (-388735), IRX5 (+4163)
HypoHier260	chr14	37641134	37641964	-0.83	7.49	17478	76696	76704	9	9	8.37E-05	2.00E-03	SLC25A21 (+522)
HypoHier259	chr22	50737978	50738890	-0.83	3.33	37624	104583	104586	4	4	1.75E-03	2.40E-02	MAPK11 (-29612), PLXNB2 (+7622)
HypoHier258	chr5	149546691	149546946	-0.83	2.50	45829	32721	32723	3	3	4.38E-03	5.40E-02	CDX1 (+461)
HypoHier257	chr11	33890131	33891205	-0.83	6.66	9912	63280	63287	8	8	1.33E-04	3.00E-03	FBXO3 (-94606), LMO2 (+23168)
HypoHier256	chr6	28504908	28505429	-0.83	3.33	47408	35633	35636	4	4	1.74E-03	2.40E-02	GPX5 (+11467), SCAND3 (+49943)
HypoHier255	chr1	43736290	43736600	-0.83	2.50	2300	3821	3823	3	3	4.38E-03	5.40E-02	TMEM125 (+749)
HypoHier254	chr17	38334161	38334623	-0.83	5.83	24869	89455	89461	7	9	2.16E-04	3.00E-03	RAPGEFL1 (+150)
HypoHier253	chr2	223164854	223164869	-0.83	2.50	34178	16669	16671	3	21	4.36E-03	5.30E-02	PAX3 (-1162)
HypoHier252	chr19	37997294	37998171	-0.84	8.37	29127	97174	97183	10	10	5.30E-05	2.00E-03	ZNF793 (-16574), ZNF570 (+37751)
HypoHier251	chr1	4714008	4714171	-0.84	2.51	508	958	960	3	3	4.32E-03	5.10E-02	AJAP1 (-1015)

HypoHier250	chr19	38307803	38308263	-0.84	3.35	29135	97206	97209	4	4	1.71E-03	2.20E-02	SIPA1L3 (-89835), ZNF573 (-43642)
HypoHier249	chr8	35092687	35092876	-0.84	3.35	54553	50100	50103	4	4	1.71E-03	2.20E-02	UNC5D (-193)
HypoHier248	chr2	206550819	206551466	-0.84	2.51	33783	15993	15995	3	4	4.30E-03	5.10E-02	NRP2 (+3919), INO80D (+399763)
HypoHier247	chr5	15499895	15500913	-0.84	5.87	43870	28928	28934	7	7	2.13E-04	3.00E-03	FBXL7 (+99)
HypoHier246	chr19	4474970	4475136	-0.84	2.52	27990	95083	95085	3	3	4.29E-03	5.10E-02	ENSG00000167674 (+2769), PLIN4 (+42663)
HypoHier245	chr11	126286370	126286828	-0.84	3.35	12276	67216	67219	4	4	1.70E-03	2.20E-02	ST3GAL4 (+10922), KIRREL3 (+584056)
HypoHier244	chr19	49249932	49250561	-0.84	5.87	29736	98258	98264	7	7	2.13E-04	3.00E-03	IZUMO1 (-81)
HypoHier243	chr1	228464049	228464232	-0.84	2.52	5456	9309	9311	3	7	4.29E-03	5.10E-02	OBSCN (+68310), TRIM11 (+130400)
HypoHier242	chr10	129535138	129535968	-0.84	9.25	8452	60187	60197	11	11	3.17E-05	2.00E-03	FOXI2 (+54)
HypoHier241	chr14	38091400	38092175	-0.84	5.89	17492	76759	76765	7	7	2.11E-04	3.00E-03	SSTR1 (-585416), FOXA1 (-27549)
HypoHier240	chr8	145910546	145911897	-0.84	6.73	56387	53315	53322	8	9	1.28E-04	3.00E-03	ARHGAP39 (-80138), ZNF251 (+69768)
HypoHier239	chr11	24518149	24518448	-0.84	7.58	9744	62891	62899	9	9	7.99E-05	2.00E-03	LUZP2 (-425)
HypoHier238	chr18	74961196	74962794	-0.84	16.84	27391	94090	94109	20	28	1.42E-06	0.00E+00	GALR1 (-510)
HypoHier237	chr1	88928168	88928467	-0.84	2.53	3072	5216	5218	3	6	4.24E-03	4.90E-02	PKN2 (-221587)
HypoHier236	chr7	4832112	4832535	-0.84	4.22	50712	42936	42940	5	5	7.32E-04	9.00E-03	AP5Z1 (+17071), PAPOLB (+69301)
HypoHier235	chr10	60935663	60937257	-0.85	11.84	6879	57299	57312	14	14	9.93E-06	0.00E+00	PHYHIPL (+110)
HypoHier234	chr7	100886961	100887364	-0.85	2.54	52362	46188	46190	3	3	4.18E-03	4.70E-02	CLDN15 (-5062), FIS1 (+1203)
HypoHier233	chr11	6947541	6947773	-0.85	4.23	9309	62053	62057	5	7	7.21E-04	9.00E-03	ZNF215 (+3)
HypoHier232	chr7	73894730	73895278	-0.85	5.09	51894	45272	45277	6	6	3.55E-04	5.00E-03	GTF2I (-177007), GTF2IRD1 (+26704)
HypoHier231	chr1	164290179	164290833	-0.85	6.78	4238	7265	7272	8	8	1.25E-04	3.00E-03	PBX1 (-238366), NUF2 (+998762)
HypoHier230	chr4	101439954	101440278	-0.85	2.54	42296	26058	26060	3	3	4.16E-03	4.60E-02	EMCN (-866)
HypoHier229	chr10	95653378	95654134	-0.85	5.93	7521	58359	58365	7	7	2.03E-04	3.00E-03	SLC35G1 (+26)
HypoHier228	chr6	105388668	105388731	-0.85	2.55	49052	39879	39881	3	5	4.15E-03	4.40E-02	HACE1 (-80906), LIN28B (-16223)
HypoHier227	chr11	69286145	69286352	-0.85	3.40	10944	65024	65027	4	4	1.63E-03	2.00E-02	CCND1 (-169606), TPCN2 (+469884)
HypoHier226	chr3	16216094	16216215	-0.85	3.41	37989	18420	18423	4	4	1.61E-03	2.00E-02	GALNT15 (-1)
HypoHier225	chr19	46318439	46319398	-0.85	7.66	29532	97906	97914	9	9	7.76E-05	2.00E-03	RSPH6A (-342)
HypoHier224	chr6	27635558	27635849	-0.85	3.41	47352	35502	35505	4	4	1.60E-03	2.00E-02	ZNF184 (-194807), HIST1H2BL (+140005)
HypoHier223	chr3	181444131	181445647	-0.85	10.25	40425	22654	22665	12	14	1.94E-05	1.00E-03	SOX2 (+15167)
HypoHier222	chr13	97646639	97647084	-0.86	6.85	16562	74878	74885	8	9	1.19E-04	3.00E-03	OXGR1 (-258)
HypoHier221	chr4	126237986	126238929	-0.86	5.14	42555	26572	26577	6	6	3.41E-04	4.00E-03	FAT4 (+904)
HypoHier220	chr2	5835853	5837348	-0.86	8.57	30551	10410	10419	10	10	4.80E-05	2.00E-03	SOX11 (+3802)
HypoHier219	chr3	164914150	164915196	-0.86	11.14	40210	22282	22294	13	16	1.25E-05	1.00E-03	SLITRK3 (-883)
HypoHier218	chr13	50706583	50707586	-0.86	6.86	16233	74277	74284	8	8	1.19E-04	3.00E-03	RNASEH2B (-776729), DLEU1 (+50778)
HypoHier217	chr8	79578116	79578360	-0.86	4.30	55236	51396	51400	5	6	6.79E-04	8.00E-03	ZC2HC1A (-44)
HypoHier216	chr8	38831148	38832728	-0.86	7.74	54657	50258	50266	9	9	7.35E-05	2.00E-03	HTRA4 (+255)
HypoHier215	chr1	95392925	95393429	-0.86	4.30	3185	5452	5456	5	5	6.77E-04	8.00E-03	CNN3 (-343)
HypoHier214	chr11	121971092	121971247	-0.86	3.45	12115	66951	66954	4	8	1.55E-03	1.90E-02	BLID (+15753), SORL1 (+648258)
HypoHier213	chr10	1651238	1651391	-0.86	2.59	5993	55747	55749	3	3	3.94E-03	4.10E-02	IDI1 (-556205), ADARB2 (+128355)
HypoHier212	chr7	26897182	26897714	-0.86	6.05	51025	43602	43608	7	7	1.91E-04	3.00E-03	SKAP2 (+6914), SNX10 (+565905)
HypoHier211	chr7	121512781	121513944	-0.87	6.06	52630	46597	46603	7	7	1.89E-04	3.00E-03	PTPRZ1 (+220)
HypoHier210	chr12	97379628	97379917	-0.87	2.60	14417	70861	70863	3	3	3.88E-03	3.60E-02	NEDD1 (+78529)
HypoHier209	chr1	1108820	1109832	-0.87	5.21	70	135	140	6	6	3.26E-04	4.00E-03	TTLL10 (+43)
HypoHier208	chr12	4918169	4919591	-0.87	7.81	12766	68003	68011	9	9	7.07E-05	2.00E-03	KCNA6 (+538)
HypoHier207	chr13	70681804	70682324	-0.87	3.48	16389	74512	74515	4	11	1.49E-03	1.50E-02	KLHL1 (+527)
HypoHier206	chr7	49812836	49813763	-0.87	7.83	51544	44664	44672	9	9	7.02E-05	2.00E-03	VWC2 (+43)
HypoHier205	chr4	6202091	6202609	-0.87	6.11	41188	24086	24092	7	7	1.84E-04	3.00E-03	JAKMIP1 (-68)
HypoHier204	chr6	161188322	161188638	-0.87	3.49	49935	41463	41466	4	4	1.48E-03	1.50E-02	MAP3K4 (-224336), PLG (+65206)
HypoHier203	chr2	241496830	241497599	-0.87	6.11	34725	17636	17642	7	7	1.83E-04	3.00E-03	DUSP28 (-2504), ANKMY1 (+165)
HypoHier202	chr2	45240373	45241008	-0.87	2.62	31380	11764	11766	3	6	3.79E-03	3.50E-02	SIX2 (-4122)
HypoHier201	chr2	154729059	154730157	-0.88	7.01	33102	14608	14615	8	9	1.11E-04	3.00E-03	KCNJ3 (-825203), GALNT13 (+1182)
HypoHier200	chr17	79140481	79140589	-0.88	2.63	26539	92363	92365	3	3	3.76E-03	3.40E-02	AATK (-718)
HypoHier199	chr20	37352612	37353158	-0.88	6.14	35445	100732	100738	7	7	1.80E-04	2.00E-03	SLC32A1 (-220)
HypoHier198	chr3	3842202	3842717	-0.88	4.38	37700	17918	17922	5	5	6.29E-04	5.00E-03	SETMAR (-502528), LRRN1 (+1339)
HypoHier197	chr2	46043	46558	-0.88	2.63	30345	10062	10064	3	3	3.75E-03	3.20E-02	FAM110C (+84)
HypoHier196	chr1	26644234	26645161	-0.88	5.27	1614	2646	2651	6	7	3.10E-04	3.00E-03	UBXN11 (+156), CD52 (+250)
HypoHier195	chr7	134576049	134576386	-0.88	5.28	52892	47096	47101	6	6	3.07E-04	3.00E-03	AGBL3 (-95041), CALD1 (+111817)
HypoHier194	chr17	17625521	17626127	-0.88	2.64	24286	88416	88418	3	3	3.72E-03	3.10E-02	RAI1 (+41037), SREBF1 (+114477)
HypoHier193	chr1	3663164	3663705	-0.88	4.41	461	894	898	5	5	6.13E-04	4.00E-03	SMIM1 (-25917), TP73 (+94351)

HypoHier192	chr7	153584416	153585368	-0.88	7.07	53274	47734	47741	8	8	1.08E-04	2.00E-03	DPP6 (-164873)
HypoHier191	chr8	110986371	110987019	-0.88	4.42	55645	52087	52091	5	5	6.07E-04	4.00E-03	SYBU (-282675), KCNV1 (+1381)
HypoHier190	chr11	118402280	118402540	-0.89	2.66	11964	66736	66738	3	3	3.65E-03	2.60E-02	TMEM25 (+486)
HypoHier189	chr2	236578808	236579780	-0.89	3.54	34470	17183	17186	4	6	1.40E-03	1.20E-02	AGAP1 (+176543), GBX2 (+497718)
HypoHier188	chr1	145714010	145714143	-0.89	2.67	3598	6202	6204	3	3	3.61E-03	2.50E-02	PDZK1 (-29186), CD160 (-7216)
HypoHier187	chr9	13278512	13278977	-0.89	2.67	56481	53484	53486	3	3	3.59E-03	2.40E-02	MPDZ (+818)
HypoHier186	chr2	179184854	179184866	-0.89	2.67	33497	15541	15543	3	3	3.59E-03	2.40E-02	OSBPL6 (+125652), PRKRA (+131098)
HypoHier185	chr6	19837015	19838211	-0.89	7.15	47165	35080	35087	8	8	1.02E-04	2.00E-03	ID4 (-4)
HypoHier184	chr13	75149833	75149974	-0.89	2.68	16410	74547	74549	3	3	3.54E-03	2.20E-02	KLF12 (-580718), TBC1D4 (+906346)
HypoHier183	chr16	1585644	1586043	-0.89	2.68	21241	83181	83183	3	3	3.54E-03	2.20E-02	TMEM204 (+2270), IFT140 (+76267)
HypoHier182	chr3	238048	238618	-0.90	7.16	37664	17849	17856	8	8	1.01E-04	2.00E-03	CHL1 (+54)
HypoHier181	chr16	54966471	54967786	-0.90	5.38	22349	84962	84967	6	6	2.91E-04	3.00E-03	IRX6 (-390543), IRX5 (+2355)
HypoHier180	chr1	155146302	155146785	-0.90	3.59	3917	6744	6747	4	4	1.33E-03	1.00E-02	KRTCAP2 (-735), TRIM46 (+101)
HypoHier179	chr2	106362547	106362832	-0.90	2.69	32366	13364	13366	3	3	3.50E-03	2.20E-02	C2orf40 (-319422), NCK2 (+1336)
HypoHier178	chr13	46425508	46426026	-0.90	8.08	16156	74148	74156	9	9	6.22E-05	2.00E-03	SIAH3 (+104)
HypoHier177	chr14	27065974	27066771	-0.90	4.49	17342	76351	76355	5	5	5.71E-04	4.00E-03	NOVA1 (+587)
HypoHier176	chr7	79081666	79084078	-0.90	20.73	51985	45392	45414	23	26	4.73E-07	0.00E+00	MAGI2 (+18)
HypoHier175	chr12	133195061	133195520	-0.90	3.61	15611	73148	73151	4	4	1.31E-03	1.00E-02	P2RX2 (-112)
HypoHier174	chr11	101453871	101454823	-0.90	8.12	11586	66087	66095	9	11	6.12E-05	2.00E-03	TRPC6 (+312)
HypoHier173	chr8	121137734	121138133	-0.90	2.71	55734	52234	52236	3	5	3.43E-03	2.10E-02	COL14A1 (+587)
HypoHier172	chr6	5851031	5851840	-0.91	3.63	46857	34497	34500	4	4	1.29E-03	1.00E-02	NRN1 (+155764), FARS2 (+590159)
HypoHier171	chr7	2756996	2758014	-0.91	3.63	50643	42788	42791	4	7	1.28E-03	1.00E-02	AMZ1 (+38349), GNA12 (+126453)
HypoHier170	chr8	57069688	57070013	-0.91	2.73	54929	50788	50790	3	4	3.37E-03	2.10E-02	MOS (-43310), PLAG1 (+53987)
HypoHier169	chr15	48469952	48470794	-0.91	6.37	19652	80363	80369	7	7	1.58E-04	2.00E-03	MYEF2 (+341)
HypoHier168	chr6	1601229	1601802	-0.91	4.55	46734	34317	34321	5	5	5.49E-04	4.00E-03	FOXC1 (-9165), FOXF2 (+211447)
HypoHier167	chr4	174421377	174422908	-0.91	7.28	42996	27293	27300	8	8	9.60E-05	2.00E-03	SCRG1 (-101456), HAND2 (+29237)
HypoHier166	chr9	125796809	125797284	-0.91	2.73	57287	54567	54569	3	3	3.35E-03	2.00E-02	GPR21 (+241)
HypoHier165	chr7	142494148	142494492	-0.91	5.47	53038	47320	47325	6	9	2.74E-04	3.00E-03	EPHB6 (-58472), PRSS1 (+36990)
HypoHier164	chr17	1617102	1617465	-0.91	4.57	23705	87292	87296	5	6	5.40E-04	4.00E-03	TLCD2 (-3552)
HypoHier163	chr1	24645802	24646392	-0.91	10.97	1531	2499	2510	12	12	1.37E-05	1.00E-03	GRHL3 (+182)
HypoHier162	chr15	41803428	41804283	-0.92	4.58	19527	80162	80166	5	5	5.38E-04	4.00E-03	LTK (+2148), ITPKA (+17783)
HypoHier161	chr5	74162621	74162924	-0.92	6.41	44494	30091	30097	7	7	1.54E-04	2.00E-03	FAM169A (-110)
HypoHier160	chr2	45159894	45160554	-0.92	7.33	31348	11672	11679	8	8	9.34E-05	2.00E-03	SIX3 (-8678), CAMKMT (+571121)
HypoHier159	chr12	96252066	96252245	-0.92	2.76	14398	70830	70832	3	6	3.25E-03	1.80E-02	SNRPF (-550)
HypoHier158	chr16	8768031	8768425	-0.92	2.76	21596	83780	83782	3	4	3.24E-03	1.80E-02	ABAT (-38598), METTL22 (+52683)
HypoHier157	chr5	178367621	178368620	-0.92	10.12	46533	33940	33950	11	12	2.08E-05	2.00E-03	ZNF454 (-71)
HypoHier156	chr4	165877875	165878219	-0.92	6.45	42933	27197	27203	7	7	1.51E-04	2.00E-03	MARCH1 (-572845), TRIM61 (+20773)
HypoHier155	chr6	33039396	33039500	-0.92	4.61	47937	37696	37700	5	5	5.24E-04	4.00E-03	HLA-DPB1 (-4255)
HypoHier154	chr4	81118188	81119473	-0.92	8.30	42106	25727	25735	9	11	5.68E-05	2.00E-03	FGF5 (-68922), PRDM8 (+12407)
HypoHier153	chr11	32454718	32455192	-0.92	7.40	9878	63212	63219	8	11	8.87E-05	2.00E-03	WT1 (+2221), RCN1 (+342505)
HypoHier152	chr12	66122804	66123496	-0.93	4.63	14043	70267	70271	5	6	5.17E-04	4.00E-03	HMGA2 (-94761), MSRB3 (+450727)
HypoHier151	chr16	54321848	54323498	-0.93	7.43	22334	84924	84931	8	8	8.63E-05	2.00E-03	IRX3 (-1998)
HypoHier150	chr4	187476326	187476953	-0.93	9.30	43229	27758	27767	10	13	2.98E-05	2.00E-03	MTNR1A (+81)
HypoHier149	chr3	42947263	42947385	-0.93	2.79	38285	18950	18952	3	7	3.12E-03	1.80E-02	ZNF662 (-334)
HypoHier148	chr19	53758289	53758609	-0.93	4.66	30029	98883	98887	5	5	5.01E-04	4.00E-03	VN1R2 (-3096), ZNF677 (-323)
HypoHier147	chr21	34405553	34405997	-0.93	3.73	36277	102259	102262	4	4	1.15E-03	8.00E-03	OLIG1 (-36675), OLIG2 (+7532)
HypoHier146	chr19	57617602	57618436	-0.93	3.73	30269	99326	99329	4	4	1.15E-03	8.00E-03	ZIM2 (-265922), USP29 (-13392)
HypoHier145	chr10	96162066	96162189	-0.94	5.61	7535	58385	58390	6	7	2.52E-04	3.00E-03	TBC1D12 (-133)
HypoHier144	chr6	31430641	31431969	-0.94	7.49	47715	36852	36859	8	8	8.37E-05	1.00E-03	MICB (-34587), MICA (+59949)
HypoHier143	chr7	751331	752456	-0.94	9.37	50352	42180	42189	10	13	2.84E-05	1.00E-03	PRKAR1B (+950)
HypoHier142	chr5	166405485	166406845	-0.94	5.63	46094	33181	33186	6	6	2.49E-04	1.00E-03	TENM2 (-305639)
HypoHier141	chr16	54315551	54316049	-0.94	3.75	22330	84911	84914	4	4	1.12E-03	7.00E-03	IRX3 (+4875), FTO (+577925)
HypoHier140	chr7	14028690	14029701	-0.94	12.22	50838	43208	43220	13	13	7.80E-06	0.00E+00	ETV1 (+375)
HypoHier139	chr17	9479910	9480428	-0.94	8.46	24107	88076	88084	9	9	5.01E-05	1.00E-03	STX8 (-616), WDR16 (+225)
HypoHier138	chr2	74875227	74875548	-0.94	7.55	31926	12652	12659	8	8	8.11E-05	1.00E-03	M1AP (-224)
HypoHier137	chr12	63327142	63327207	-0.94	2.83	14003	70204	70206	3	3	3.00E-03	1.60E-02	PPM1H (+1642), MON2 (+466578)
HypoHier136	chr1	60280088	60281077	-0.94	8.50	2747	4603	4611	9	9	5.01E-05	1.00E-03	HOOK1 (+50)
HypoHier135	chr2	241459638	241460002	-0.94	2.83	34724	17633	17635	3	3	2.99E-03	1.60E-02	ANKMY1 (+37560), GPC1 (+84732)

HypoHier134	chr11	65194933	65195039	-0.95	3.78	10672	64518	64521	4	4	1.09E-03	7.00E-03	SCYL1 (-97562), FRMD8 (+40916)
HypoHier133	chr12	71833017	71833193	-0.95	3.79	14133	70386	70389	4	4	1.09E-03	7.00E-03	LGR5 (-445)
HypoHier132	chr8	688018	688417	-0.95	2.84	53689	48568	48570	3	3	2.95E-03	1.60E-02	DLGAP2 (-761314), TDRP (-192437)
HypoHier131	chr9	115818857	115819364	-0.95	3.80	57158	54384	54387	4	5	1.07E-03	7.00E-03	ZFP37 (-115)
HypoHier130	chr6	30458158	30458730	-0.95	6.65	47620	36515	36521	7	7	1.35E-04	1.00E-03	PRR3 (-66219), HLA-E (+1200)
HypoHier129	chr19	863054	863244	-0.95	3.81	27604	94453	94456	4	4	1.06E-03	7.00E-03	CFD (+3696), MED16 (+30069)
HypoHier128	chr8	674434	674560	-0.95	2.86	53687	48561	48563	3	3	2.90E-03	1.60E-02	DLGAP2 (-775035), TDRP (-178716)
HypoHier127	chr11	125774082	125774447	-0.96	5.75	12258	67189	67194	6	6	2.31E-04	1.00E-03	PUS3 (-1149), DDX25 (+7)
HypoHier126	chr1	209404846	209405064	-0.97	2.91	5023	8582	8584	3	3	2.75E-03	1.20E-02	PLXNA2 (-987290), CAMK1G (-352107)
HypoHier125	chr11	40314618	40315404	-0.97	4.85	9990	63393	63397	5	5	4.28E-04	1.00E-03	LRRC4C (+653)
HypoHier124	chr8	54163622	54164442	-0.98	9.76	54868	50654	50663	10	11	2.46E-05	0.00E+00	OPRK1 (+225)
HypoHier123	chr5	140345643	140346403	-0.98	10.75	45491	31960	31970	11	12	1.49E-05	0.00E+00	PCDHAC2 (+203)
HypoHier122	chr2	240302585	240302855	-0.98	3.91	34665	17544	17547	4	4	9.71E-04	6.00E-03	HDAC4 (+19923), TWIST2 (+546047)
HypoHier121	chr1	228644750	228646224	-0.98	18.60	5466	9333	9351	19	19	9.46E-07	0.00E+00	HIST3H2BB (-321), HIST3H2A (+73)
HypoHier120	chr5	150284416	150284796	-0.98	6.86	45867	32787	32793	7	7	1.19E-04	0.00E+00	ZNF300 (-61)
HypoHier119	chr2	154727925	154728468	-0.98	5.89	33101	14602	14607	6	7	2.10E-04	0.00E+00	GALNT13 (-229)
HypoHier118	chr7	98990265	98990837	-0.98	2.94	52235	45974	45976	3	3	2.64E-03	1.20E-02	BUD31 (-15713), ARPC1B (+18198)
HypoHier117	chr7	155599026	155599780	-0.98	3.94	53364	47956	47959	4	4	9.45E-04	6.00E-03	SHH (+5564), RBM33 (+162030)
HypoHier116	chr6	33091357	33091841	-0.98	5.91	47947	37760	37765	6	6	2.06E-04	0.00E+00	HLA-DPA1 (-43112), COL11A2 (+68677)
HypoHier115	chr4	110223040	110224247	-0.99	15.76	42371	26170	26185	16	16	1.66E-06	0.00E+00	COL25A1 (+155)
HypoHier114	chr3	128204047	128205496	-0.99	8.88	39646	21186	21194	9	9	4.04E-05	0.00E+00	DNAJB8 (-18961), GATA2 (+7256)
HypoHier113	chr6	41605343	41605784	-0.99	2.96	48317	38450	38452	3	3	2.59E-03	1.20E-02	MDFI (-621)
HypoHier112	chr18	20713759	20714332	-0.99	2.96	26971	93224	93226	3	6	2.58E-03	1.20E-02	CABLES1 (-1681)
HypoHier111	chr4	467983	468201	-0.99	5.93	40811	23307	23312	6	6	2.04E-04	0.00E+00	ZNF721 (+24853), ZNF141 (+136468)
HypoHier110	chr7	1067302	1067459	-0.99	2.97	50402	42278	42280	3	3	2.57E-03	1.20E-02	GPR146 (-27540), CYP2W1 (+44546)
HypoHier109	chr6	10529935	10530035	-0.99	2.97	46982	34781	34783	3	3	2.57E-03	1.20E-02	GCNT6 (-104008), GCNT2 (+1396)
HypoHier108	chr9	90113813	90114156	-0.99	3.97	56854	53976	53979	4	4	9.23E-04	6.00E-03	CTSL (-226449), DAPK1 (+1189)
HypoHier107	chr15	35046760	35047411	-0.99	5.96	19363	79897	79902	6	8	2.01E-04	0.00E+00	GJD2 (+80)
HypoHier106	chr1	144930572	144931379	-0.99	2.98	3573	6170	6172	3	3	2.54E-03	1.20E-02	PDE4DIP (+64046), NBPF9 (+119228)
HypoHier105	chr9	136149908	136150823	-0.99	4.97	57639	55017	55021	5	5	3.86E-04	1.00E-03	OBP2B (-65738), SURF6 (+52869)
HypoHier104	chr3	16974160	16974681	-1.00	2.99	38001	18436	18438	3	3	2.50E-03	1.20E-02	PLCL2 (+47969), TBC1D5 (+767091)
HypoHier103	chr1	228646916	228646970	-1.00	2.99	5467	9352	9354	3	3	2.50E-03	1.20E-02	HIST3H2A (-1383), HIST3H2BB (+1135)
HypoHier102	chr4	103998289	103998497	-1.00	5.01	42314	26078	26082	5	5	3.77E-04	1.00E-03	SLC9B2 (-463)
HypoHier101	chr16	17562960	17563300	-1.00	3.01	21749	84007	84009	3	3	2.44E-03	1.10E-02	XYLT1 (+1608)
HypoHier100	chr7	29923339	29924165	-1.01	7.07	51152	44037	44043	7	7	1.08E-04	0.00E+00	WIPF3 (+49411), SCRN1 (+106153)
HypoHier99	chr5	178322737	178322924	-1.01	6.09	46531	33932	33937	6	6	1.85E-04	0.00E+00	ZFP2 (-64)
HypoHier98	chr19	56988398	56989543	-1.02	7.16	30244	99262	99268	7	7	1.01E-04	0.00E+00	ZNF667 (-272)
HypoHier97	chr19	19221316	19221574	-1.02	4.09	28785	96511	96514	4	4	8.19E-04	2.00E-03	TMEM161A (+27857), SLC25A42 (+46637)
HypoHier96	chr11	124747075	124747263	-1.02	4.10	12220	67126	67129	4	4	8.14E-04	2.00E-03	ROBO3 (+11887), ROBO4 (+21008)
HypoHier95	chr19	11199851	11199965	-1.02	6.15	28345	95712	95717	6	6	1.79E-04	0.00E+00	LDLR (-130)
HypoHier94	chr12	51786038	51786547	-1.03	3.09	13558	69316	69318	3	3	2.26E-03	4.00E-03	SLC4A8 (-32262), GALNT6 (-8946)
HypoHier93	chr2	164592561	164593498	-1.04	9.33	33209	14847	14855	9	11	2.93E-05	0.00E+00	FIGN (-513)
HypoHier92	chr12	62583974	62584454	-1.04	3.12	13993	70191	70193	3	3	2.19E-03	3.00E-03	FAM19A2 (+2409)
HypoHier91	chr19	51196754	51197463	-1.04	3.12	29885	98580	98582	3	3	2.19E-03	3.00E-03	SYT3 (-55807), SHANK1 (+23086)
HypoHier90	chr11	8190152	8190837	-1.05	6.27	9372	62167	62172	6	6	1.67E-04	0.00E+00	RIC3 (+41)
HypoHier89	chr17	75955921	75956222	-1.05	3.14	26263	91865	91867	3	3	2.13E-03	3.00E-03	TNRC6C (-45065), SEPT9 (+678580)
HypoHier88	chr14	24640947	24642317	-1.05	14.72	17300	76250	76263	14	14	2.60E-06	0.00E+00	REC8 (+570)
HypoHier87	chr6	152011103	152011666	-1.05	5.26	49757	41156	41160	5	5	3.11E-04	0.00E+00	ESR1 (-246)
HypoHier86	chr14	91866325	91866751	-1.06	4.25	18350	78136	78139	4	4	7.10E-04	0.00E+00	GPR68 (-146269), CCDC88C (+17583)
HypoHier85	chr2	241936844	241937034	-1.06	3.19	34762	17711	17713	3	3	2.03E-03	2.00E-03	SNED1 (-1316)
HypoHier84	chr3	44753946	44754587	-1.07	11.76	38325	19014	19024	11	11	1.04E-05	0.00E+00	ZNF502 (+129)
HypoHier83	chr14	106329158	106329652	-1.07	4.29	19024	79341	79344	4	4	6.86E-04	0.00E+00	TMEM121 (+336465)
HypoHier82	chr19	58570419	58570995	-1.08	8.61	30313	99434	99441	8	8	4.75E-05	0.00E+00	ZNF135 (+100)
HypoHier81	chr2	216946201	216946429	-1.08	3.23	33908	16202	16204	3	3	1.93E-03	1.00E-03	TMEM169 (-274), PECR (+224)
HypoHier80	chr4	6201080	6201536	-1.08	3.24	41187	24083	24085	3	5	1.92E-03	1.00E-03	JAKMIP1 (+974)
HypoHier79	chr17	37024020	37024760	-1.08	6.48	24785	89260	89265	6	6	1.47E-04	0.00E+00	LASP1 (-1722)
HypoHier78	chr3	128209772	128210541	-1.08	4.33	39649	21199	21202	4	4	6.61E-04	0.00E+00	DNAJB8 (-24346), GATA2 (+1871)
HypoHier77	chr3	185911316	185912486	-1.09	8.71	40543	22826	22833	8	8	4.47E-05	0.00E+00	ETV5 (-85000), DGKG (+168125)

HypoHier76	chr6	32908466	32909003	-1.09	8.73	47925	37655	37662	8	13	4.47E-05	0.00E+00	HLA-DMB (+112)
HypoHier75	chr19	51189671	51190179	-1.09	4.37	29884	98576	98579	4	4	6.38E-04	0.00E+00	SYT3 (-48623), SHANK1 (+30270)
HypoHier74	chr11	32448293	32450692	-1.10	15.39	9875	63190	63203	14	14	1.66E-06	0.00E+00	WT1 (+7683), RCN1 (+337043)
HypoHier73	chr10	121300857	121301343	-1.10	3.31	8177	59708	59710	3	3	1.79E-03	1.00E-03	RGS10 (+1120), GRK5 (+333999)
HypoHier72	chr17	38084119	38084459	-1.11	4.43	24849	89423	89426	4	4	6.05E-04	0.00E+00	ORMDL3 (-1195)
HypoHier71	chr4	89978251	89978566	-1.11	3.34	42214	25906	25908	3	3	1.74E-03	1.00E-03	FAM13A (-86)
HypoHier70	chr1	3309864	3310911	-1.11	5.57	409	774	778	5	5	2.59E-04	0.00E+00	ARHGEF16 (-60602), PRDM16 (+324613)
HypoHier69	chr16	54324187	54325929	-1.11	10.03	22335	84932	84940	9	9	2.20E-05	0.00E+00	IRX3 (-4383)
HypoHier68	chr6	21667333	21668049	-1.12	4.46	47181	35112	35115	4	4	5.88E-04	0.00E+00	SOX4 (+73719), PRL (+630039)
HypoHier67	chr4	114681314	114681880	-1.13	5.63	42444	26372	26376	5	5	2.49E-04	0.00E+00	CAMK2D (+627)
HypoHier66	chr11	65324768	65325249	-1.13	3.39	10685	64555	64557	3	3	1.65E-03	1.00E-03	LTBP3 (+690)
HypoHier65	chr1	47696212	47696701	-1.13	4.52	2451	4069	4072	4	4	5.62E-04	0.00E+00	TAL1 (-1014)
HypoHier64	chr7	810958	811390	-1.13	3.40	50356	42198	42200	3	3	1.62E-03	1.00E-03	SUN1 (-60964), HEATR2 (+44836)
HypoHier63	chr6	97285662	97285776	-1.14	3.41	48944	39558	39560	3	4	1.61E-03	1.00E-03	GPR63 (-380)
HypoHier62	chr21	27944586	27945708	-1.14	5.68	36186	102103	102107	5	5	2.41E-04	0.00E+00	CYYR1 (+456)
HypoHier61	chr10	128993810	128995192	-1.14	12.52	8439	60157	60167	11	11	6.86E-06	0.00E+00	NPS (-353112), DOCK1 (+400523)
HypoHier60	chr7	121784249	121784596	-1.15	5.74	52638	46614	46618	5	5	2.31E-04	0.00E+00	AASS (-10547), FEZF1 (+160136)
HypoHier59	chr17	5000803	5001281	-1.16	4.62	23892	87621	87624	4	4	5.19E-04	0.00E+00	USP6 (-18691), ZFP3 (+19499)
HypoHier58	chr3	239188	240139	-1.16	8.10	37665	17857	17863	7	7	6.15E-05	0.00E+00	CNTN6 (-894596), CHL1 (+1385)
HypoHier57	chr7	8011594	8012104	-1.17	3.50	50800	43115	43117	3	3	1.47E-03	1.00E-03	GLCCI1 (+3424), ICA1 (+290062)
HypoHier56	chr19	57183016	57183268	-1.17	3.50	30253	99287	99289	3	3	1.47E-03	1.00E-03	ZNF835 (-14)
HypoHier55	chr11	32451777	32452839	-1.17	8.22	9876	63204	63210	7	7	5.79E-05	0.00E+00	WT1 (+4868), RCN1 (+339858)
HypoHier54	chr16	10912331	10912718	-1.18	5.88	21640	83855	83859	5	5	2.12E-04	0.00E+00	TVP23A (+117)
HypoHier53	chr11	101785507	101785806	-1.18	10.62	11592	66103	66111	9	9	1.54E-05	0.00E+00	KIAA1377 (-89)
HypoHier52	chr20	44035850	44036136	-1.19	3.57	35556	100965	100967	3	3	1.36E-03	0.00E+00	DBNDD2 (-654)
HypoHier51	chr3	151178727	151178904	-1.20	3.61	40060	22002	22004	3	3	1.30E-03	0.00E+00	IGSF10 (-2319)
HypoHier50	chr8	11057947	11059303	-1.21	13.27	54015	49102	49112	11	11	5.20E-06	0.00E+00	XKR6 (+250)
HypoHier49	chr19	12911875	12912252	-1.21	3.62	28421	95872	95874	3	3	1.29E-03	0.00E+00	PRDX2 (+609)
HypoHier48	chr2	37551024	37551108	-1.21	3.62	31201	11420	11422	3	3	1.29E-03	0.00E+00	QPCT (-20651), PRKD3 (-6029)
HypoHier47	chr19	58545001	58545837	-1.22	13.38	30309	99415	99425	11	11	5.20E-06	0.00E+00	ZSCAN1 (-15)
HypoHier46	chr8	116679763	116680127	-1.22	4.88	55672	52137	52140	4	4	4.17E-04	0.00E+00	TRPS1 (+1283)
HypoHier45	chr11	6439806	6440065	-1.23	3.69	9295	62029	62031	3	3	1.21E-03	0.00E+00	APBB1 (+364)
HypoHier44	chr12	48592635	48592818	-1.24	3.71	13430	69123	69125	3	3	1.18E-03	0.00E+00	OR10AD1 (+4443), C12orf68 (+15361)
HypoHier43	chr1	208084640	208085455	-1.24	6.19	5004	8555	8559	5	5	1.76E-04	0.00E+00	CD34 (-301)
HypoHier42	chr5	137071523	137071799	-1.24	4.98	45331	31663	31666	4	4	3.84E-04	0.00E+00	KLHL3 (+118)
HypoHier41	chr1	205781722	205782099	-1.25	3.74	4923	8431	8433	3	3	1.14E-03	0.00E+00	SLC41A1 (+393)
HypoHier40	chr1	110254709	110254896	-1.25	6.24	3333	5706	5710	5	5	1.72E-04	0.00E+00	GSTM5 (-74)
HypoHier39	chr15	89920348	89922071	-1.25	17.53	20616	81963	81976	14	18	1.42E-06	0.00E+00	POLG (-43132), RHCG (+118634)
HypoHier38	chr20	3051954	3052692	-1.26	13.82	34941	99696	99706	11	11	4.26E-06	0.00E+00	OXT (+57)
HypoHier37	chr12	93966060	93967711	-1.27	10.13	14357	70752	70759	8	8	2.08E-05	0.00E+00	CRADD (-104607), SOCS2 (+3296)
HypoHier36	chr10	74034644	74034667	-1.27	3.82	7095	57676	57678	3	3	1.06E-03	0.00E+00	DDIT4 (+978)
HypoHier35	chr5	178487310	178487826	-1.28	10.23	46541	33967	33974	8	8	1.94E-05	0.00E+00	ZNF354C (+152)
HypoHier34	chr5	178003633	178004685	-1.30	6.48	46520	33913	33917	5	5	1.47E-04	0.00E+00	PHYKPL (-344373), COL23A1 (+13397)
HypoHier33	chr19	38747174	38747378	-1.32	6.62	29146	97228	97232	5	8	1.36E-04	0.00E+00	PPP1R14A (-45)
HypoHier32	chr1	208083913	208084071	-1.33	4.00	5003	8552	8554	3	3	8.94E-04	0.00E+00	CD34 (+755)
HypoHier31	chr19	12305553	12306497	-1.34	6.71	28396	95824	95828	5	5	1.30E-04	0.00E+00	ZNF136 (+32146), ZNF44 (+99603)
HypoHier30	chr1	47694517	47695475	-1.36	8.15	2450	4063	4068	6	6	6.08E-05	0.00E+00	TAL1 (+447)
HypoHier29	chr10	124133785	124133822	-1.36	4.08	8253	59823	59825	3	3	8.27E-04	0.00E+00	PLEKHA1 (-408)
HypoHier28	chr3	100053070	100053188	-1.39	4.17	39197	20521	20523	3	3	7.60E-04	0.00E+00	NIT2 (-416)
HypoHier27	chr5	76248637	76248923	-1.39	4.18	44537	30163	30165	3	3	7.59E-04	0.00E+00	CRHBP (+242)
HypoHier26	chr7	143582146	143582630	-1.39	5.57	53062	47365	47368	4	4	2.58E-04	0.00E+00	CTAGE6 (-127599), OR2F2 (-49871)
HypoHier25	chr3	57994941	57995180	-1.40	4.21	38748	19815	19817	3	3	7.40E-04	0.00E+00	FLNB (+934)
HypoHier24	chr7	65196614	65196881	-1.44	4.33	51761	45073	45075	3	3	6.57E-04	0.00E+00	VKORC1L1 (-141506), ZNF92 (+358036)
HypoHier23	chr19	58220080	58220837	-1.46	16.09	30299	99376	99386	11	11	1.66E-06	0.00E+00	ZNF154 (+77)
HypoHier22	chr2	85810844	85812023	-1.50	10.47	32052	12858	12864	7	8	1.70E-05	0.00E+00	VAMP5 (-97)
HypoHier21	chr11	32458656	32461240	-1.50	24.08	9880	63224	63239	16	16	4.73E-07	0.00E+00	WT1 (-2772)
HypoHier20	chr5	67583598	67584380	-1.52	16.72	44386	29849	29859	11	11	1.42E-06	0.00E+00	SLC30A5 (-805484), PIK3R1 (+72441)
HypoHier19	chr12	122018770	122019525	-1.54	13.83	15064	72156	72164	9	9	4.02E-06	0.00E+00	KDM2B (-259)

HypoHier18	chr8	42356637	42356871	-1.54	4.61	54742	50427	50429	3	3	5.23E-04	0.00E+00	SMIM19 (-39544), VDAC3 (+107333)
HypoHier17	chr7	2847517	2847575	-1.54	4.62	50654	42829	42831	3	3	5.19E-04	0.00E+00	GNA12 (+36412), AMZ1 (+128390)
HypoHier16	chr1	42383598	42384647	-1.55	13.97	2260	3756	3764	9	9	3.07E-06	0.00E+00	HIVEP3 (+46)
HypoHier15	chr20	11898478	11899258	-1.57	7.86	35043	99896	99900	5	5	6.93E-05	0.00E+00	NONE
HypoHier14	chr9	135039836	135040296	-1.65	4.95	57583	54943	54945	3	3	3.91E-04	0.00E+00	NTNG2 (+2732), SETX (+190306)
HypoHier13	chr7	150211761	150211939	-1.66	8.29	53146	47515	47519	5	5	5.68E-05	0.00E+00	GIMAP7 (-68)
HypoHier12	chr6	10521452	10521715	-1.68	6.72	46980	34776	34779	4	5	1.29E-04	0.00E+00	TFAP2A (-106114), GCNT2 (-7005)
HypoHier11	chr22	38201496	38201848	-1.72	6.87	37166	103806	103809	4	4	1.19E-04	0.00E+00	GCAT (-2240), H1F0 (+558)
HypoHier10	chr16	1583810	1584516	-1.72	13.79	21240	83173	83180	8	8	4.26E-06	0.00E+00	TMEM204 (+589)
HypoHier9	chr10	114712695	114713187	-1.80	7.21	8007	59325	59328	4	6	9.86E-05	0.00E+00	HABP2 (-599844), TCF7L2 (+2932)
HypoHier8	chr19	58399865	58400559	-1.83	10.96	30303	99390	99395	6	6	1.37E-05	0.00E+00	ZNF814 (+193)
HypoHier7	chr5	76249502	76250527	-1.85	9.26	44538	30166	30170	5	5	3.10E-05	0.00E+00	AGGF1 (-76195), CRHBP (+1477)
HypoHier6	chr13	76334583	76334866	-1.89	7.57	16421	74560	74563	4	4	8.02E-05	0.00E+00	LMO7 (-72)
HypoHier5	chr14	74179674	74180217	-1.90	5.69	18029	77643	77645	3	3	2.40E-04	0.00E+00	PNMA1 (+1182), ACOT6 (+96398)
HypoHier4	chr7	143579665	143580147	-1.92	7.69	53061	47361	47364	4	4	7.50E-05	0.00E+00	CTAGE6 (-125117), OR2F2 (-52353)
HypoHier3	chr11	128391271	128391494	-2.03	6.09	12325	67281	67283	3	3	1.85E-04	0.00E+00	ETS1 (+66054)
HypoHier2	chr5	76115672	76116535	-2.15	15.08	44534	30154	30160	7	7	1.89E-06	0.00E+00	S100Z (-29820), F2RL1 (+1302)
HypoHier1	chr3	194868750	194868843	-2.16	6.49	40719	23140	23142	3	4	1.46E-04	0.00E+00	LSG1 (-475591), XXYLT1 (+123099)